

Fig. 1

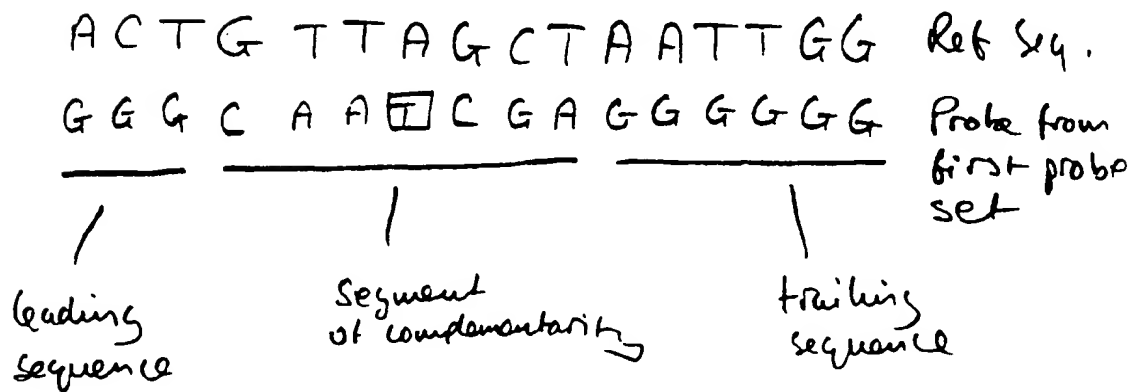


Fig. 2

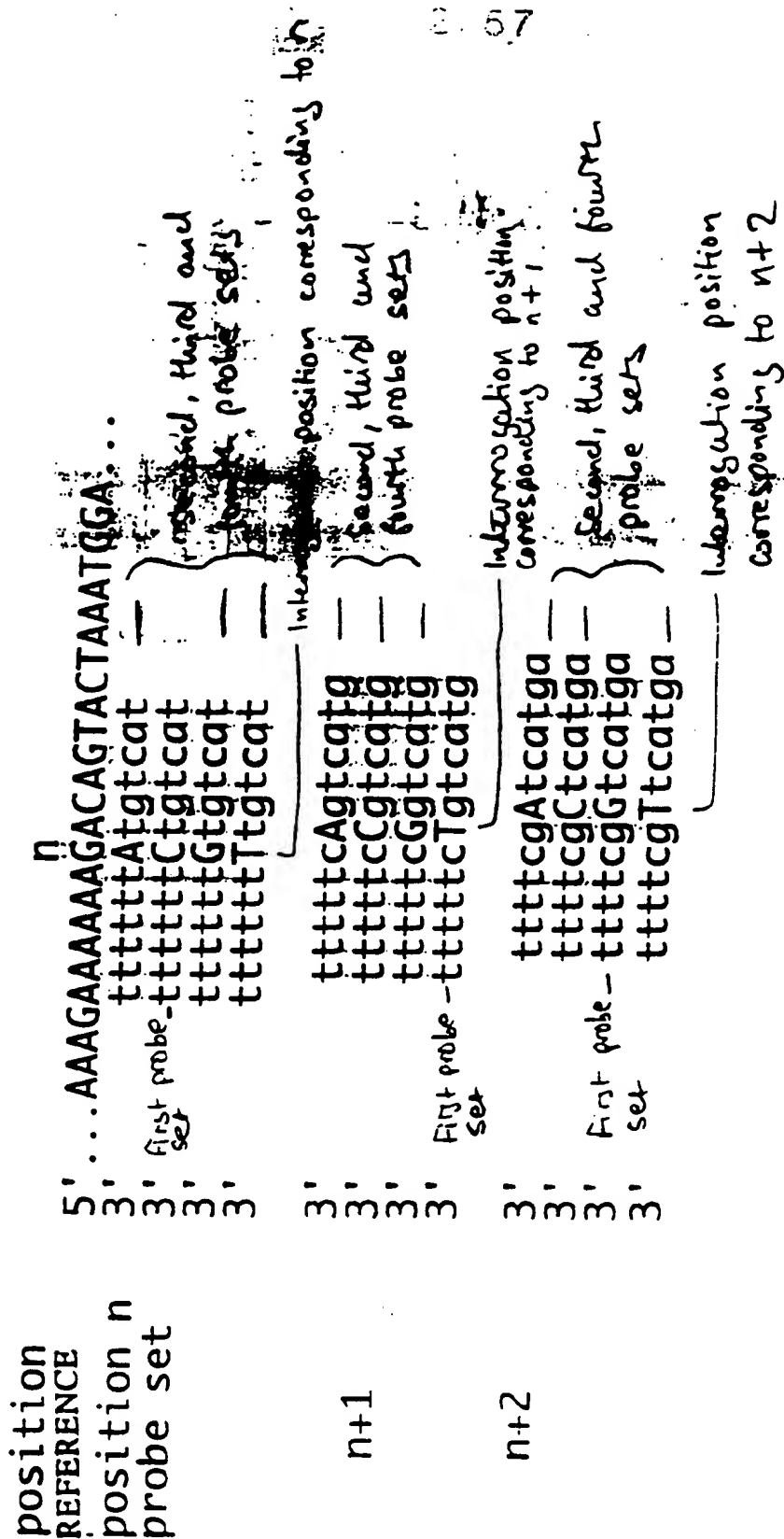


Fig. 3

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Fig. 5

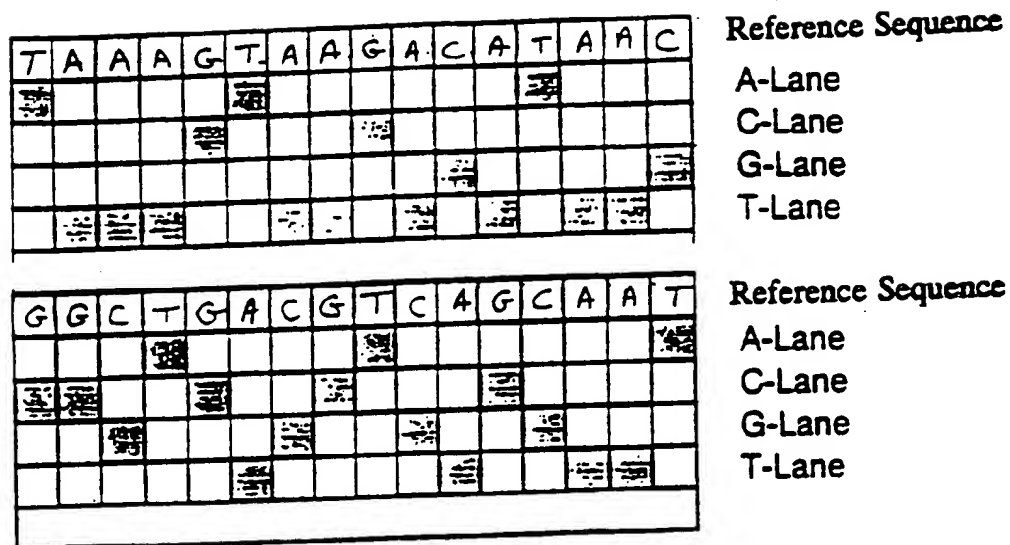


FIG. 5 : Tiled Array with Probes for the Detection of Point Mutations

3' - CCGACTACAGTCGTT
 3' - CCGACTCCAGTCGTT
 3' - CCGACTGCAGTCGTT
 3' - CCGACTTCAGTCGTT

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A C T G T T A G C T A A T T G G Ret. Seq.
 n corresponding nucleotide
 C A A T C G A — Probe from first set
 C A A — C G A T — Deletion probe
 C A A T A C G [A] } Insertion
 C A A T C C G [A] } Probes
 C A A T G C G [A] }
 C A A T T C G [A] }

Fig. 6

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α_1 α_2 α_3 Corresponding nucleotides
 A C T G T T A G C T A A T T G G Ref. Seq.

C \boxed{A} A \boxed{T} C \boxed{G} A Probe from first set
 I_1 I_2 I_3 Interrogation positions

C \boxed{C} A T C G A
 C \boxed{G} A T C G A
 C \boxed{T} A T C G A
 I_1

} Corresponding probes from second, third and fourth probe sets

C A A \boxed{A} C G A
 C A A \boxed{C} C G A
 C A A \boxed{G} C G A
 I_2

} Corresponding probes from fifth, sixth and seventh probe sets

C A A T C \boxed{A} A
 C A A T C \boxed{C} A
 C A A T C \boxed{T} A
 I_3

} Corresponding probes from eighth, ninth and tenth probe sets

Fig. 7

7157

AC^{n₃}T^{n₄}G^{n₁}TTAG^{n₂}CTAATTGG Ref. Seq.

C A **A** T C **A** A T
C A **C** T C **C** A T
C A **G** T C **G** A T
C A **T** T C **T** A T

F_1 I_2 Interrogation position

T G A C T A T
T G C C G A T
T G G C C A T
T G T C F A T

$I_3 \quad I_4$ Interrogation positions

Fig. 8

A corresponding nucleotide
ATTC CGGGATC

AGG G C C A T — Probe from first probe set
 AGG C C C A T
 AGG A C C A T
 AGG T C C A T

} corresponding probes from second, third and fourth probe sets

heine: mutation

Interruption
section

Fig. 9

3157

HV 407A

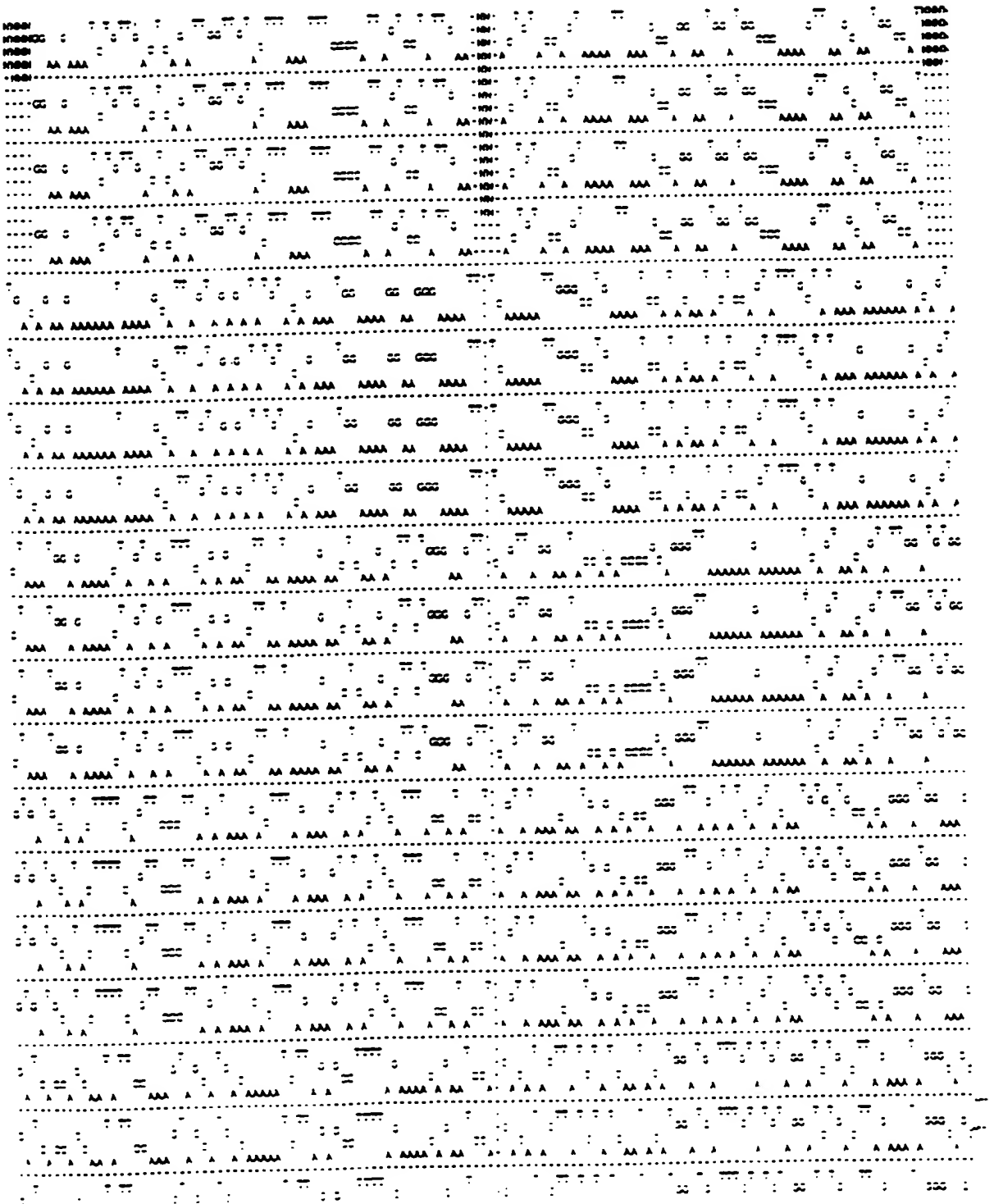
130 x 140

13/7

15/9

17/9

19/10



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HV787A (2)

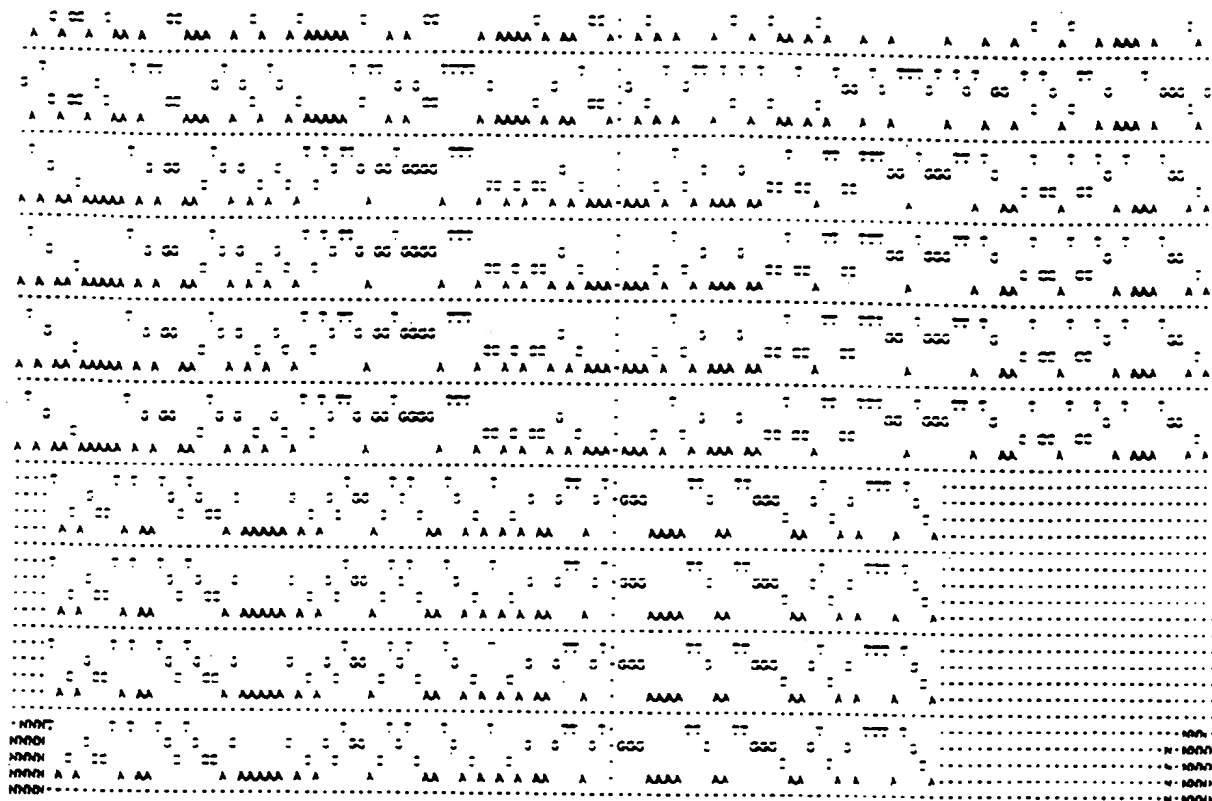
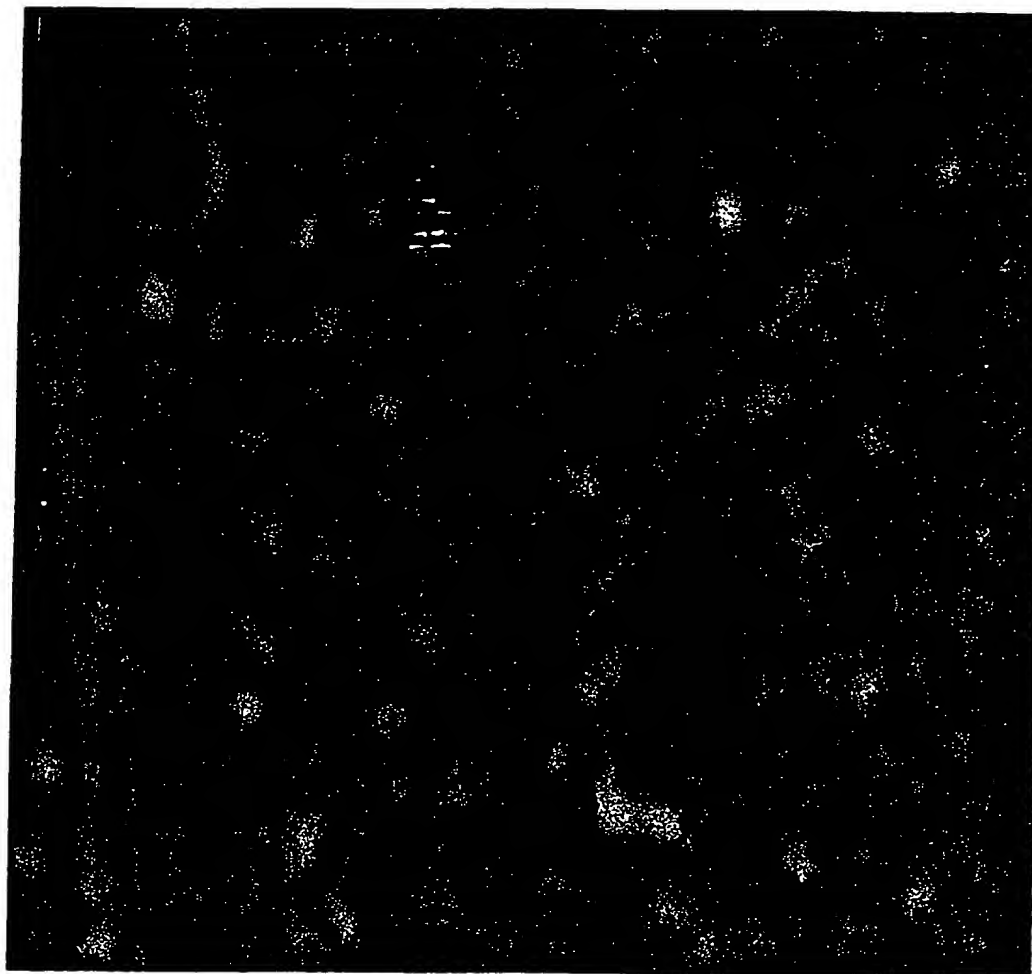


Fig. 10
Page 2 of 2

10/57



13 probe length
15
17
19

MC07060:

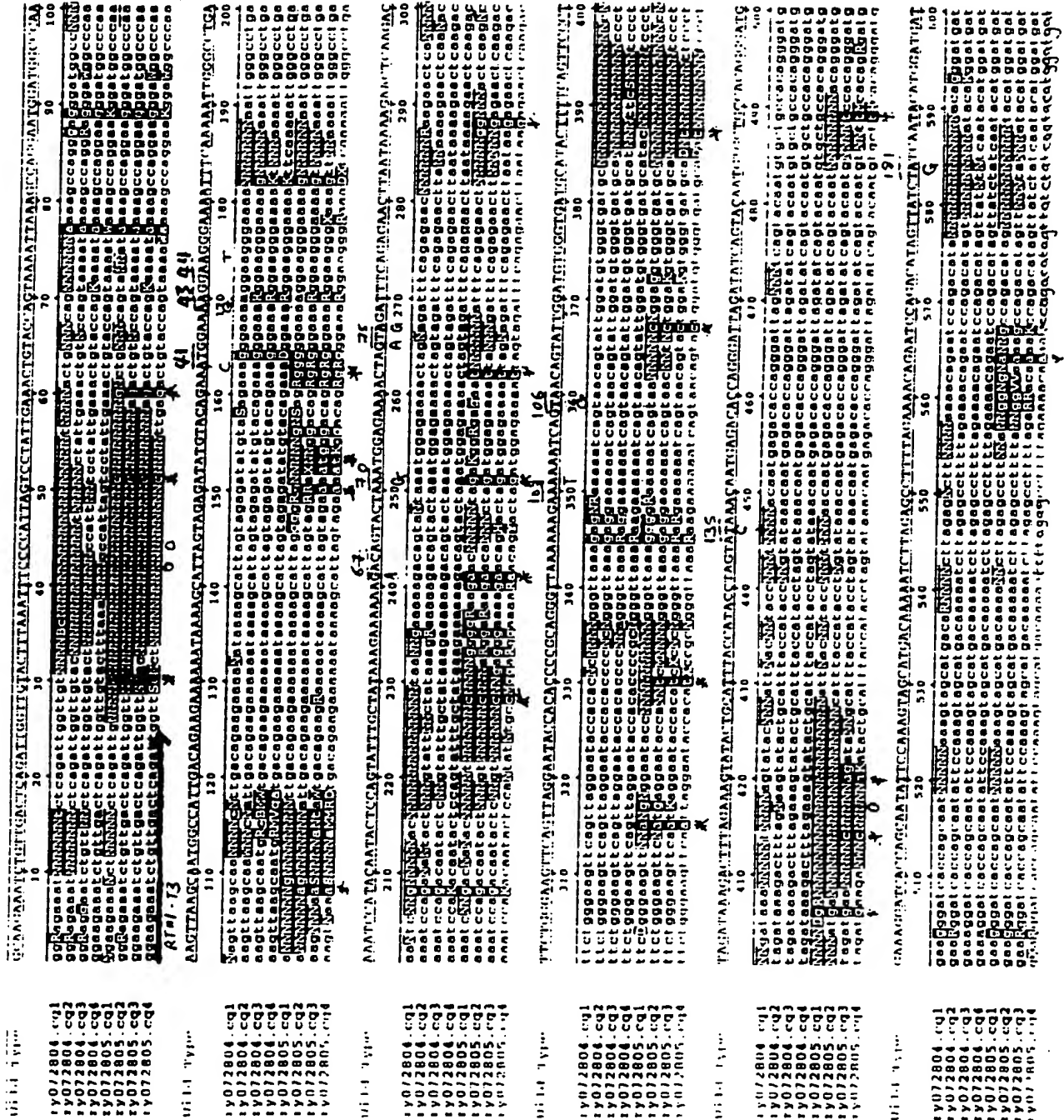
= 407 water chip hybridized with fragmented pfol 19 RNA

Fig. 11

11/57

Figure 12
(Page 1 of 2)

SF2 target: SF12 chip
4MUT18
SF2 target: SF2 chip



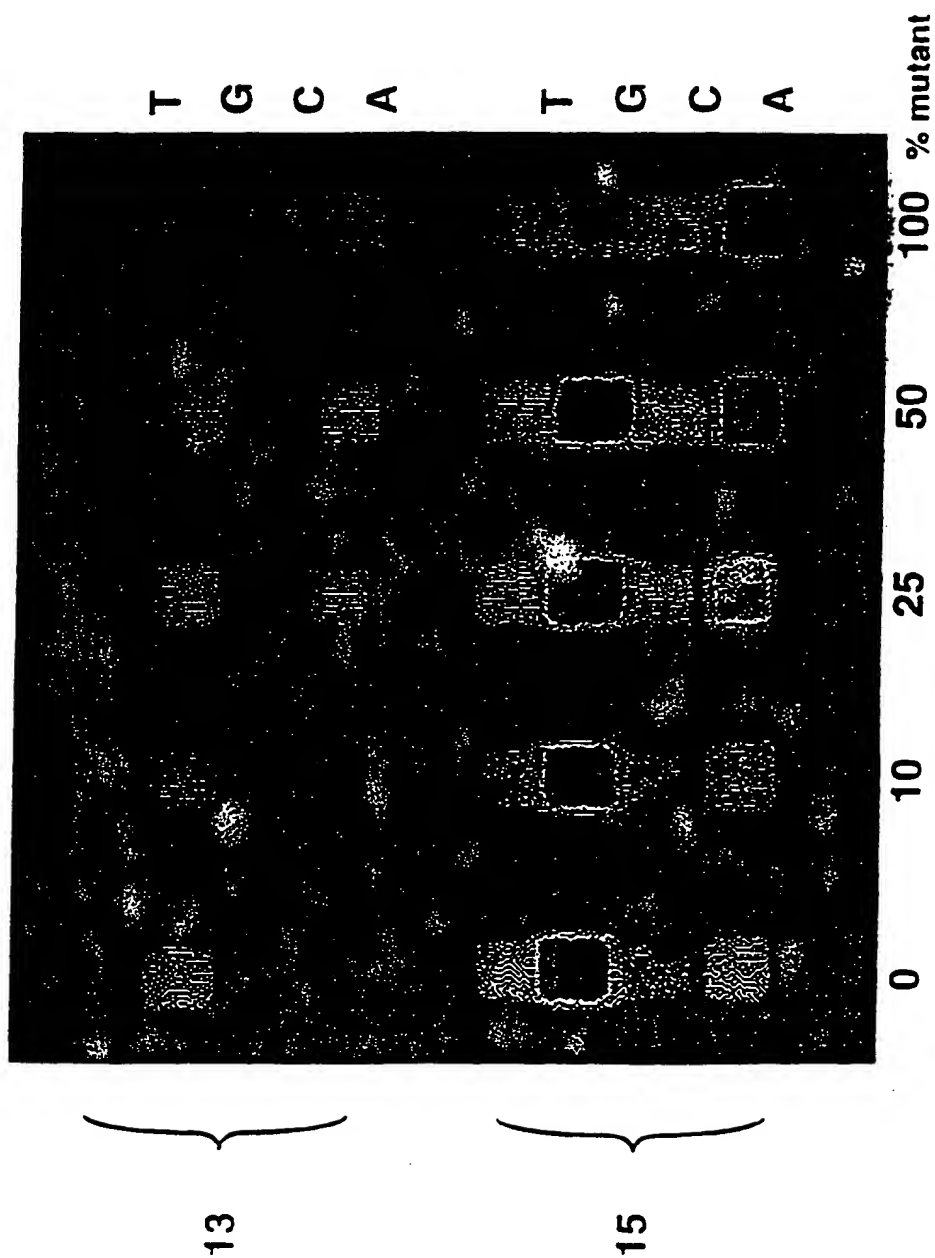
13/57

5'Fluorescein-AAAGAAAAAAGACAGTACTAAATGGAGAAAAT wildtype
 PROBE 3' tttttt•tgtcat 13mers
 PROBE 3' cttttttt•tgtcatg 15mers
 PROBE 3' tctttttt•tgtcatga 17mers
 PROBE 3' ttctttttt•tgtcatgat 19mers
 5'Fluorescein-AAAGAAAAAACAAGTACTAAATGGAGAAAAT mutant

Fig. 13

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Fig. 14



15. 57

14 pre and post- α -DDI treated Patients

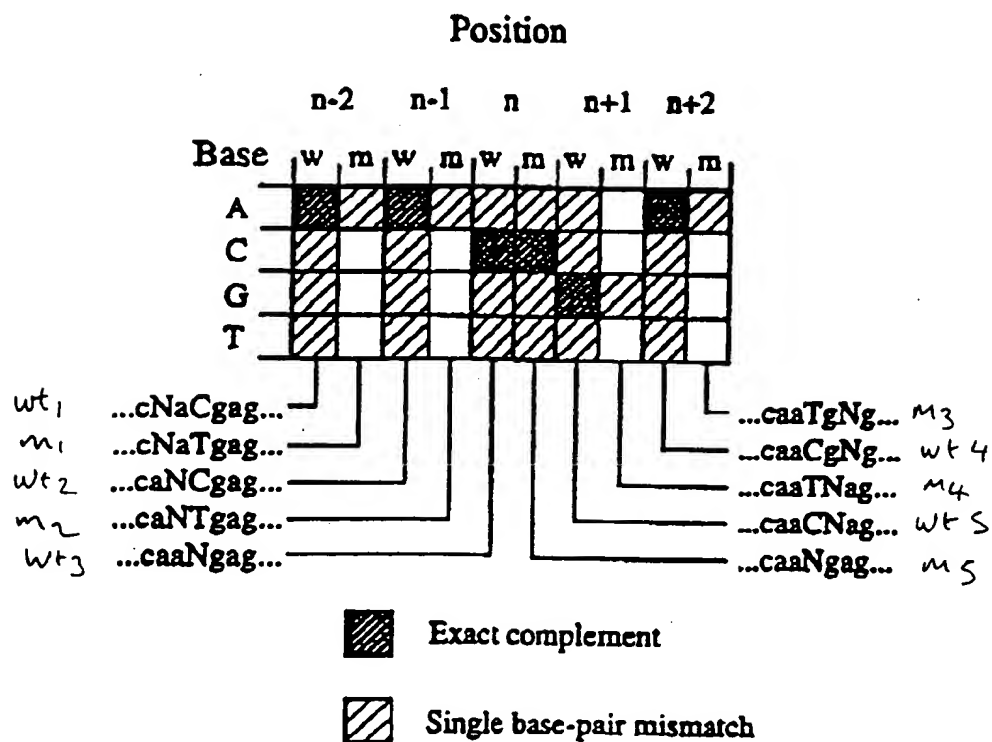
[illegible][illegible]

Fig. 15

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Array Design for the R553X Point Mutation

Wild-Type Pattern



Wild-Type Sequence: 5'-AGGTCAA**C**GAGCAA-3'

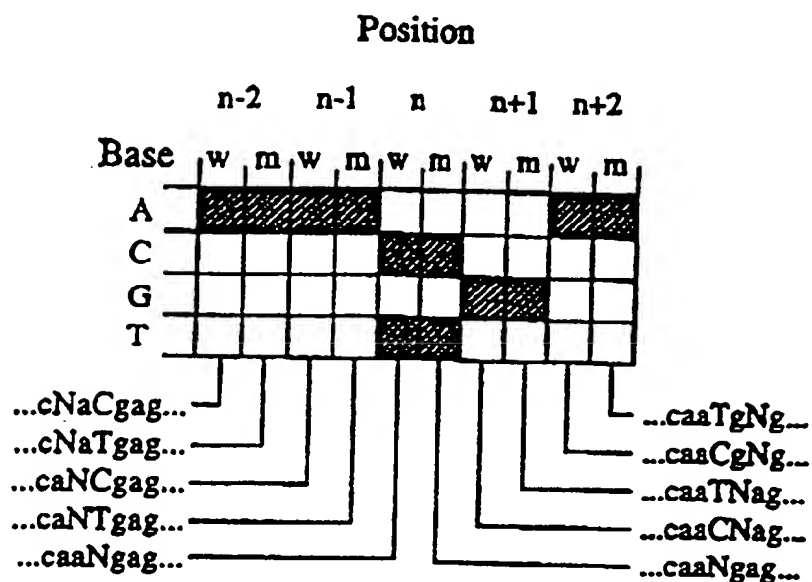
Mutant Sequence: 5'-AGGTCAA**T**GAGCAA-3'

Fig. 16

17/57

Array Design for the R553X Point Mutation

Heterozygote Pattern



Wild-Type Sequence: 5'-AGGTCAA**C**GAGCAA-3'

Mutant Sequence: 5'-AGGTCAA**T**GAGCAA-3'

Fig. 17

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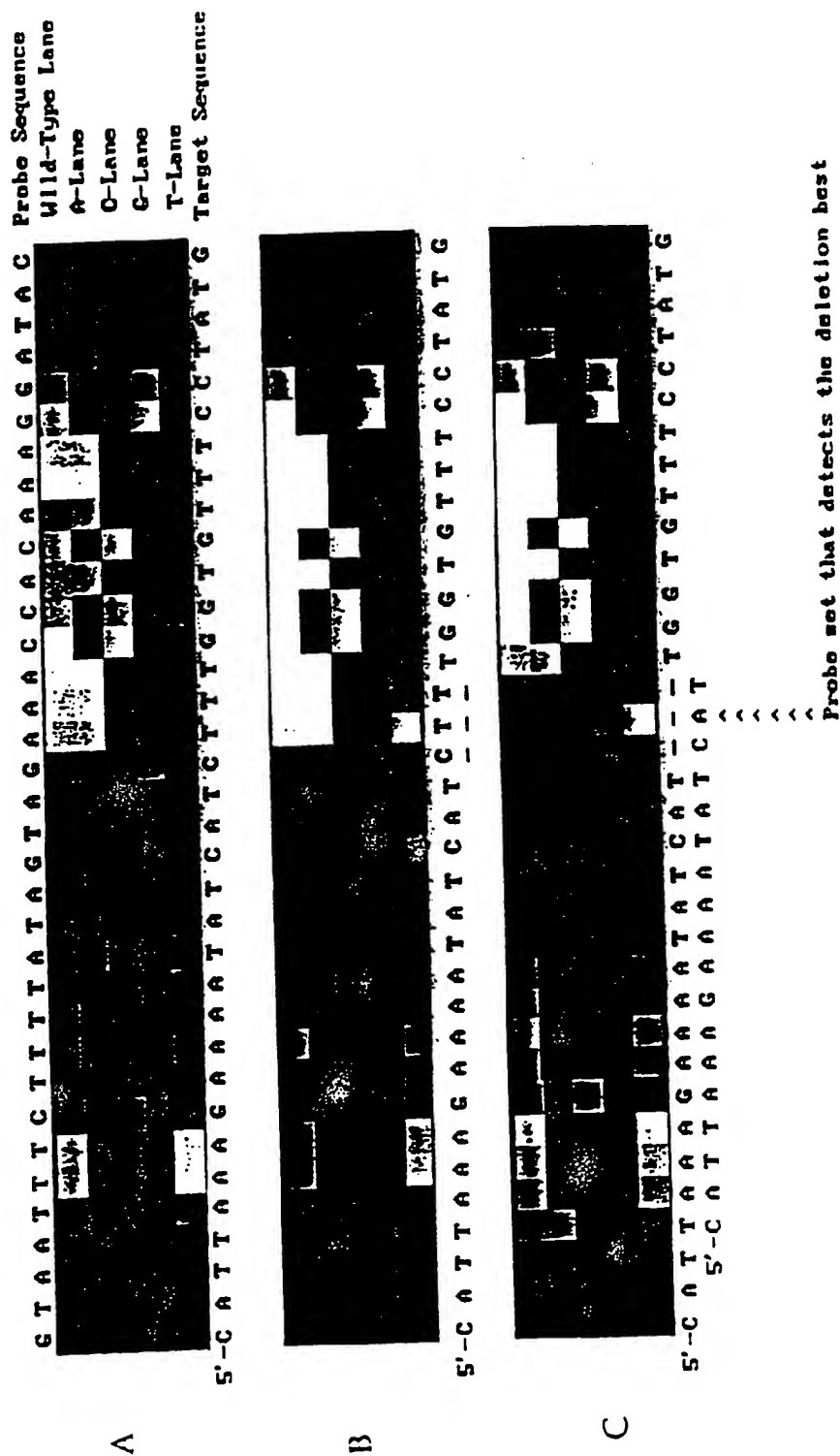


Fig. 18

19/5

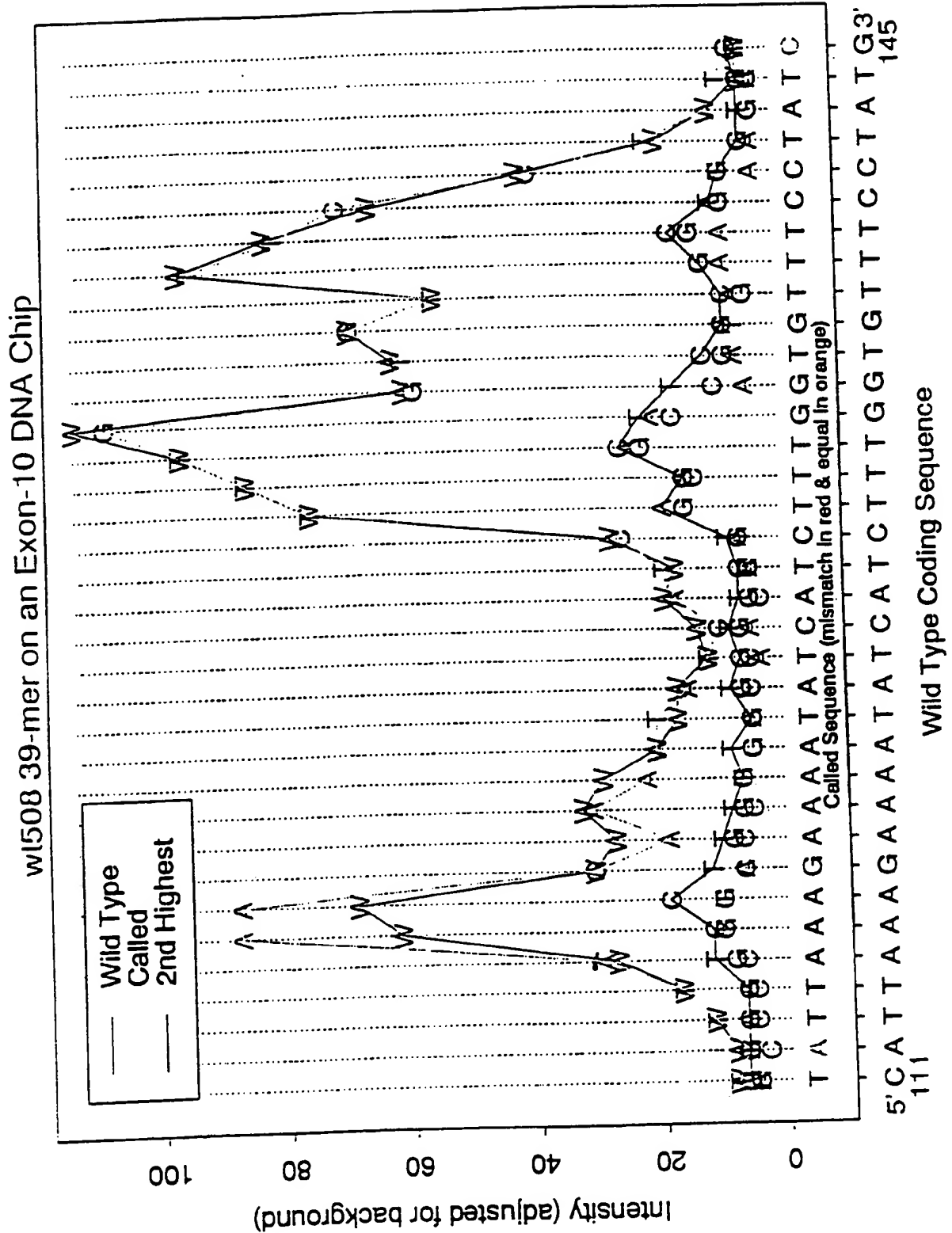


Fig. 19
Page 1 of 3

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wl508 and mu508 on an Exon-10 DNA Chip

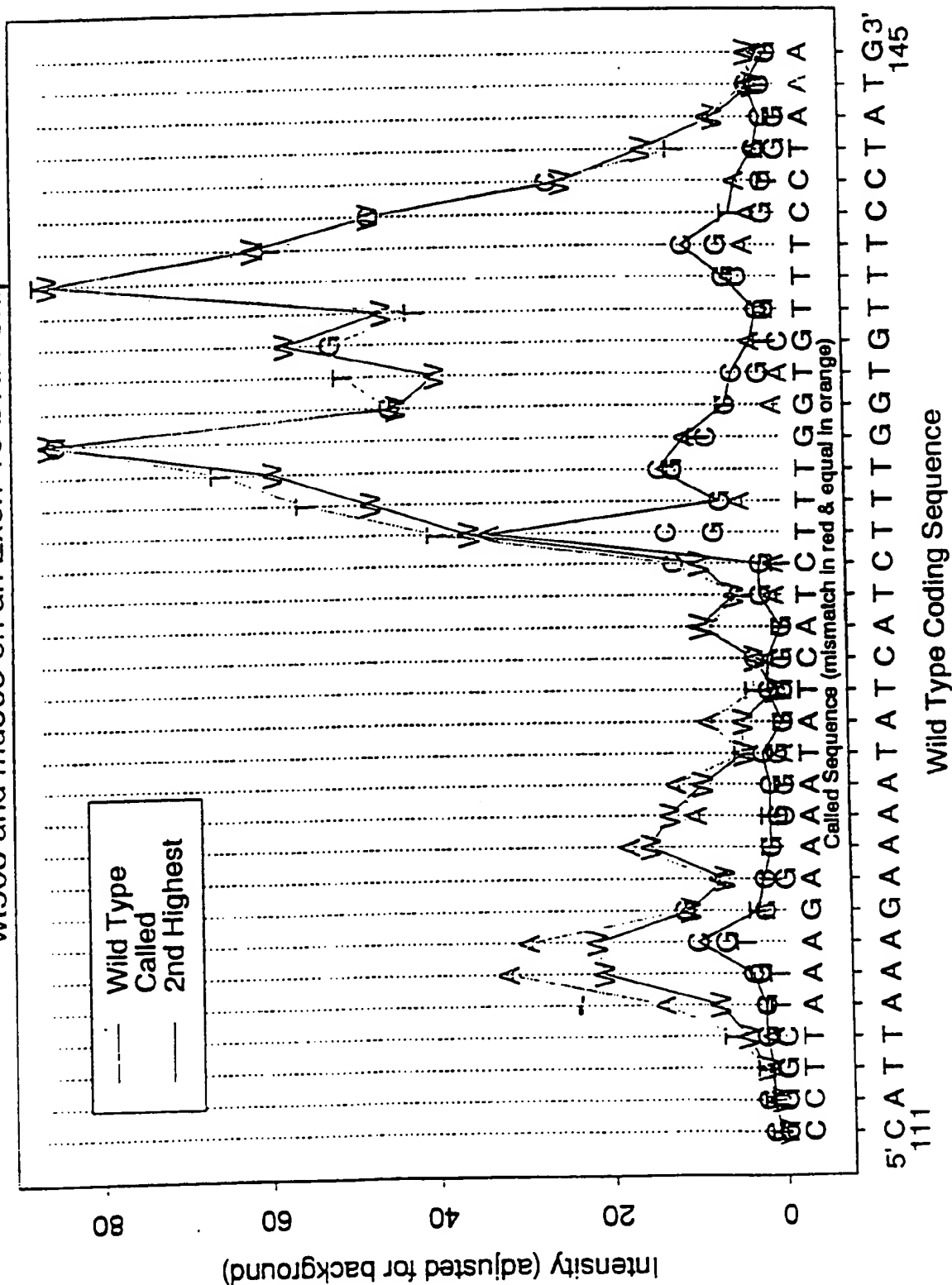


Fig. 19
Page 2 of 3

21,57

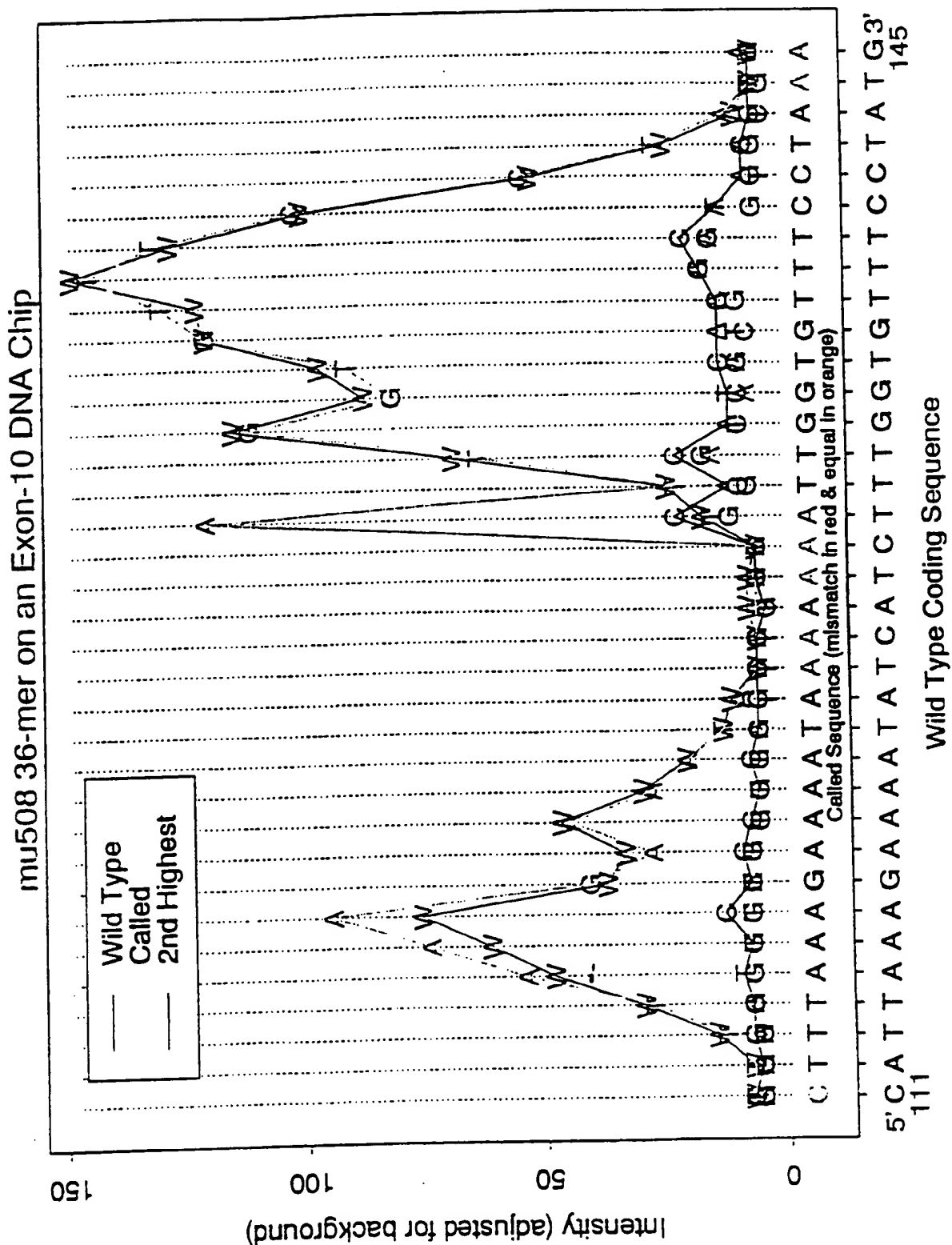


Fig. 19
Page 3 of 3

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Probe Sequence
Wild-Type Lane
A-Lane
C-Lane
G-Lane
T-Lane
Target Sequence

GGAGTCTCCCATTTAATT
5'-CCTTCAGAGGGTAATAATTAA

A

5'-CCTTCAGAGGGTAATAATTAA

B

5'-CCTTCAGAGTGTAAATAATTAA

C

Fig. 20

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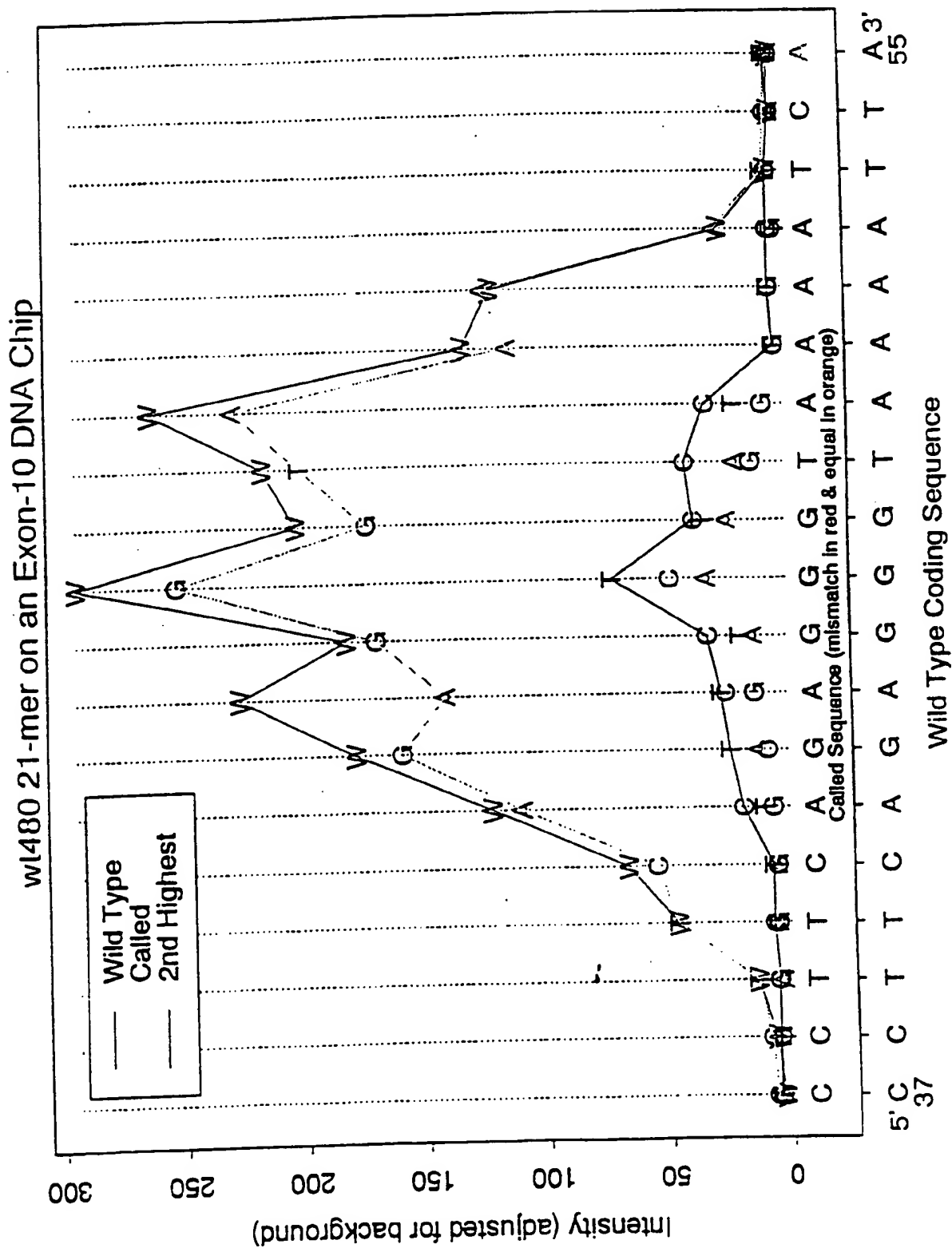


Fig. 21
Page 1 of 3

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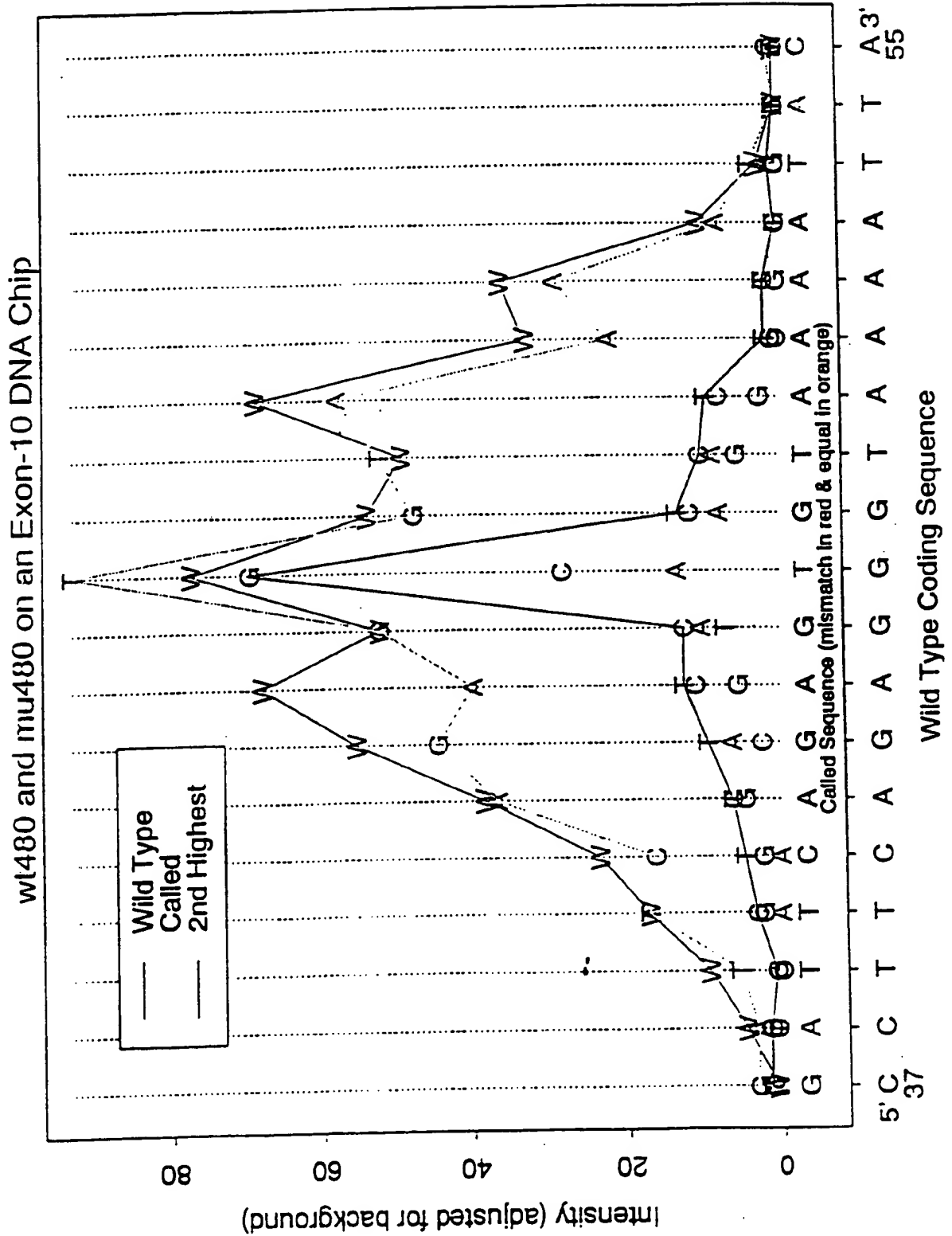


Fig. 21
Page 2 of 3

25/57 .

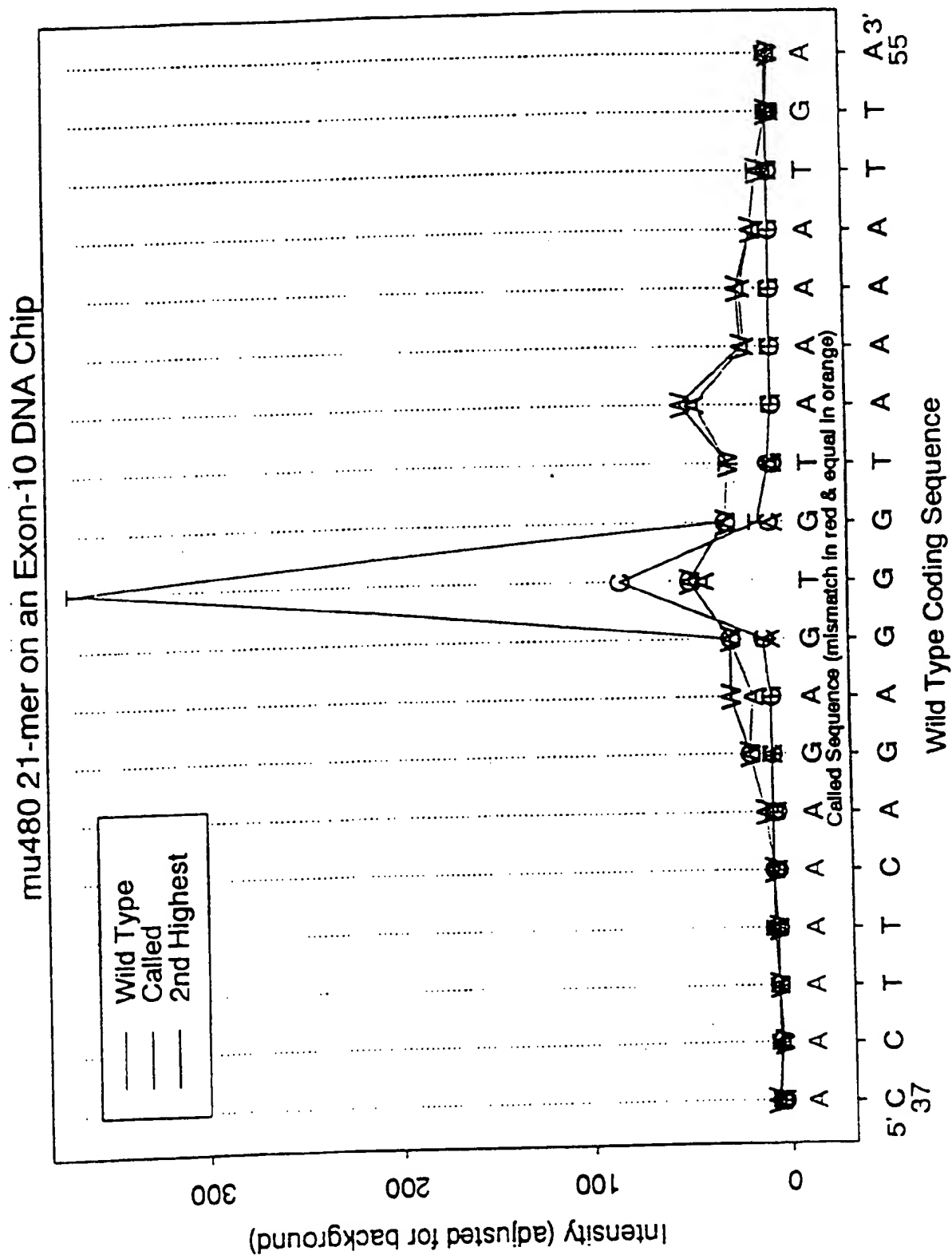


Fig. 21
Page 3 of 3

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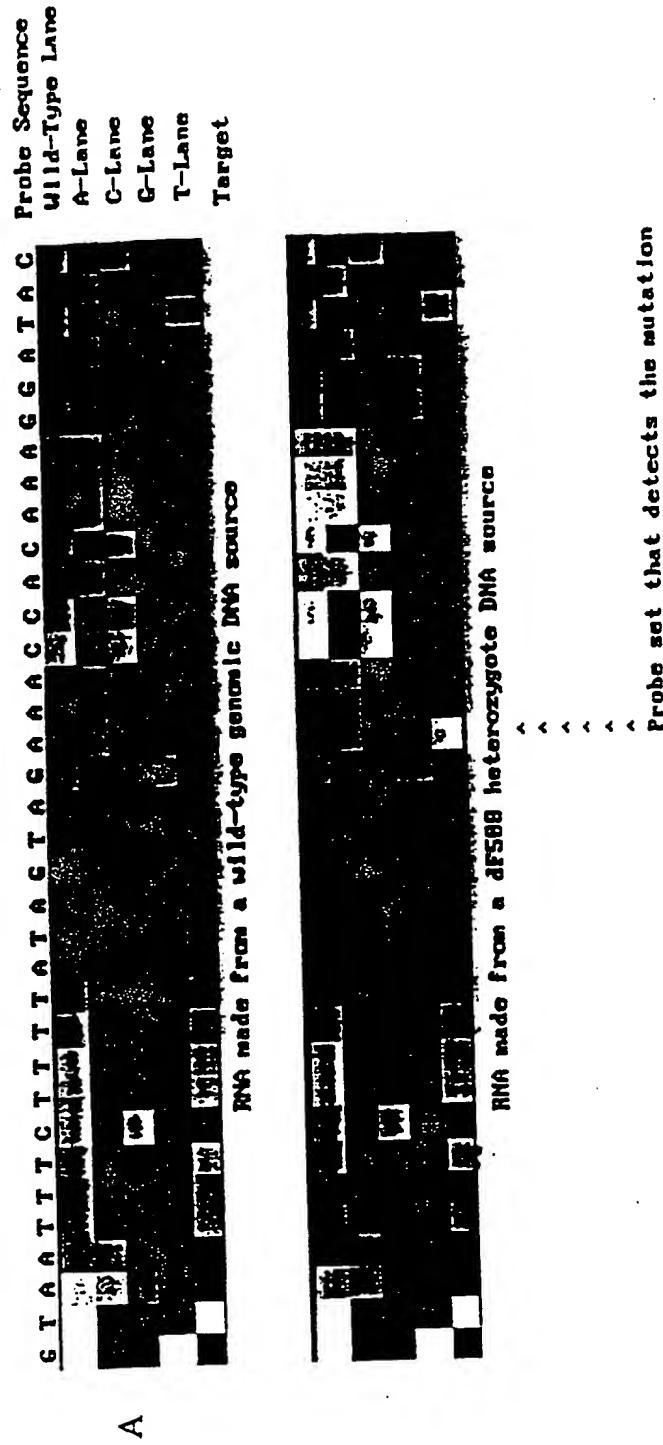


Fig. 22

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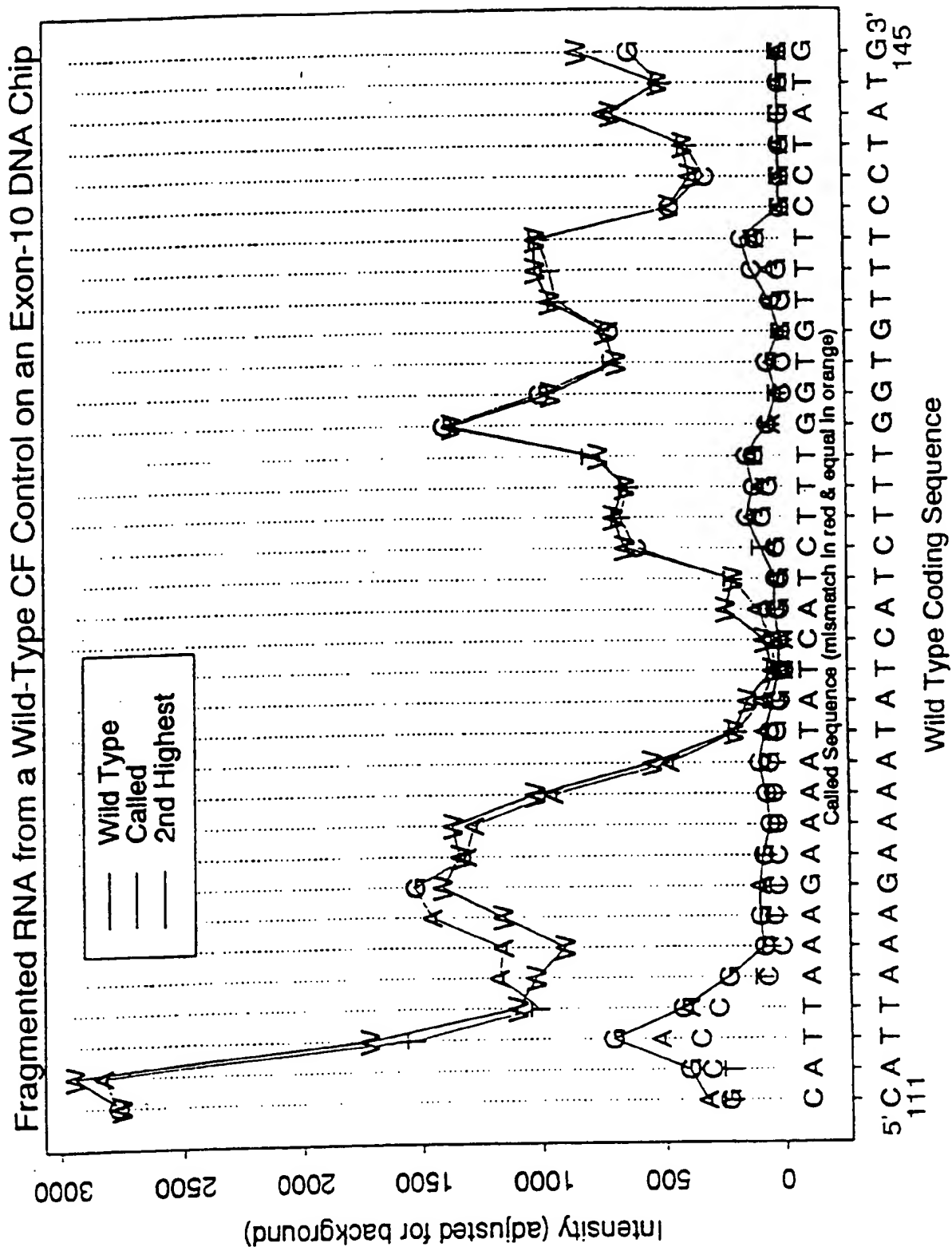


Fig. 23
Page 1 of 2

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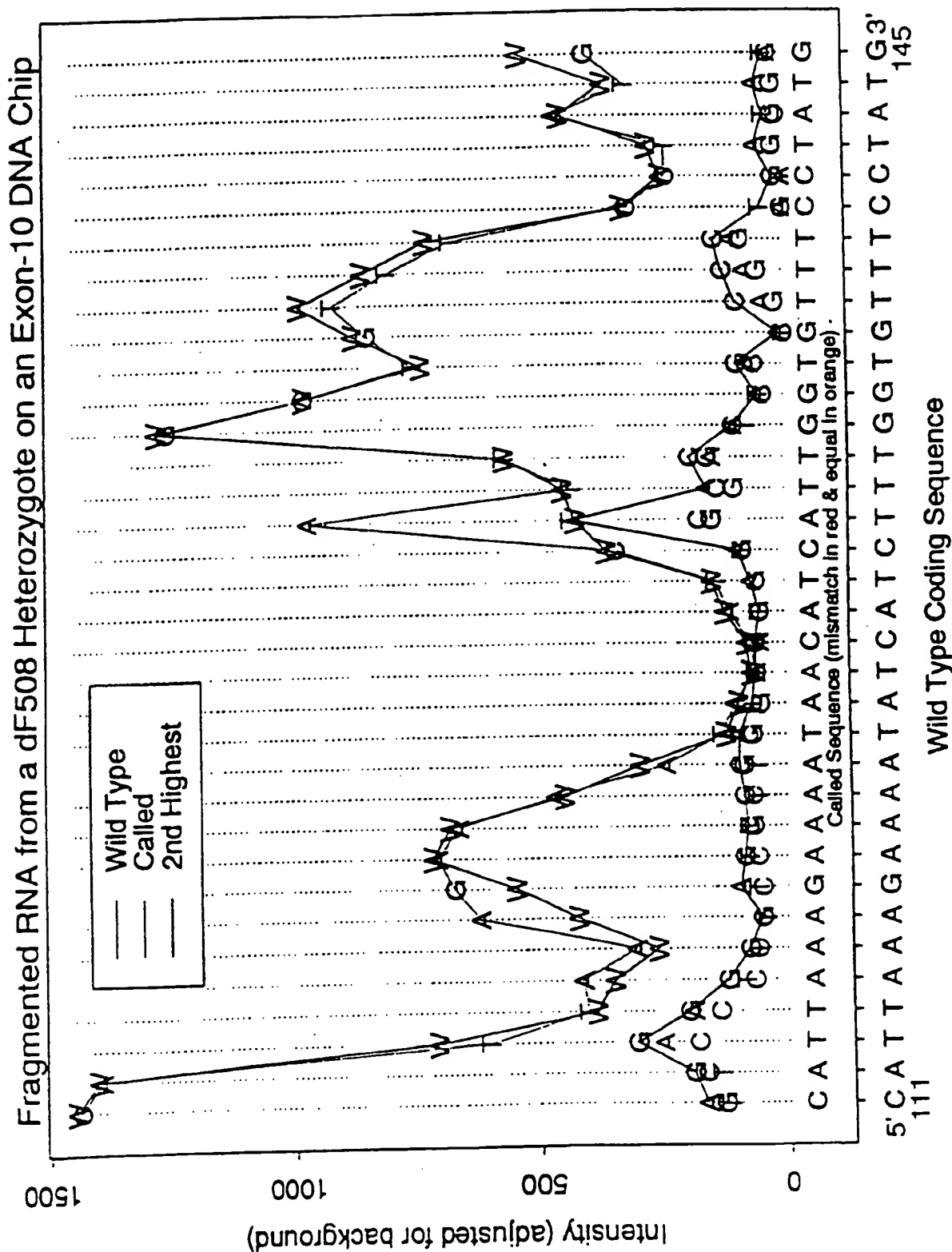


Fig. 23

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A

B

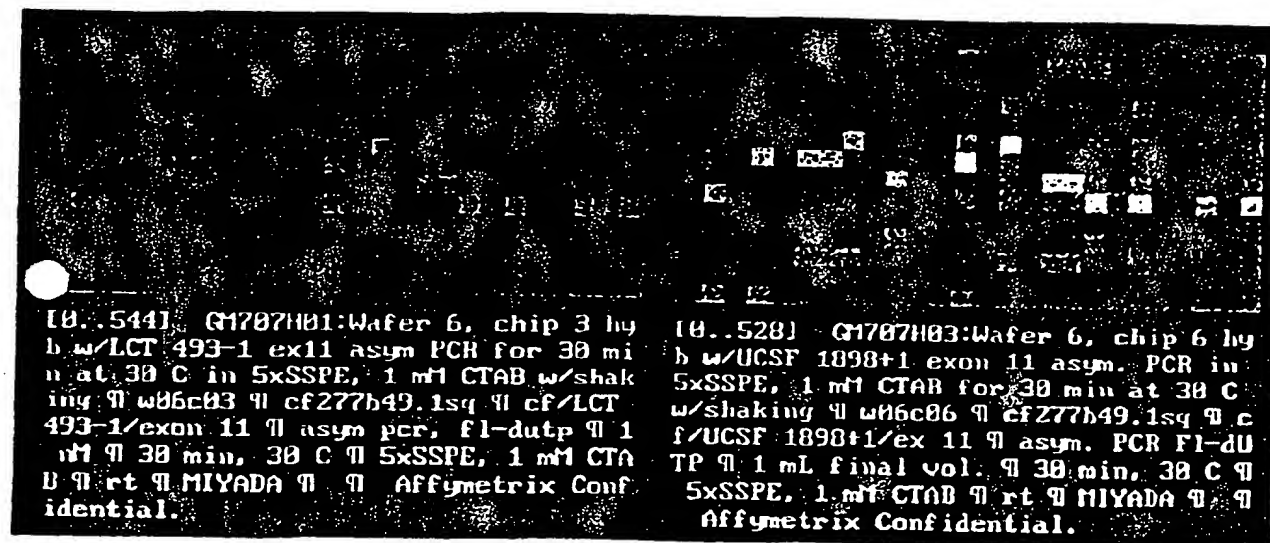


Fig. 24

30. 17

A

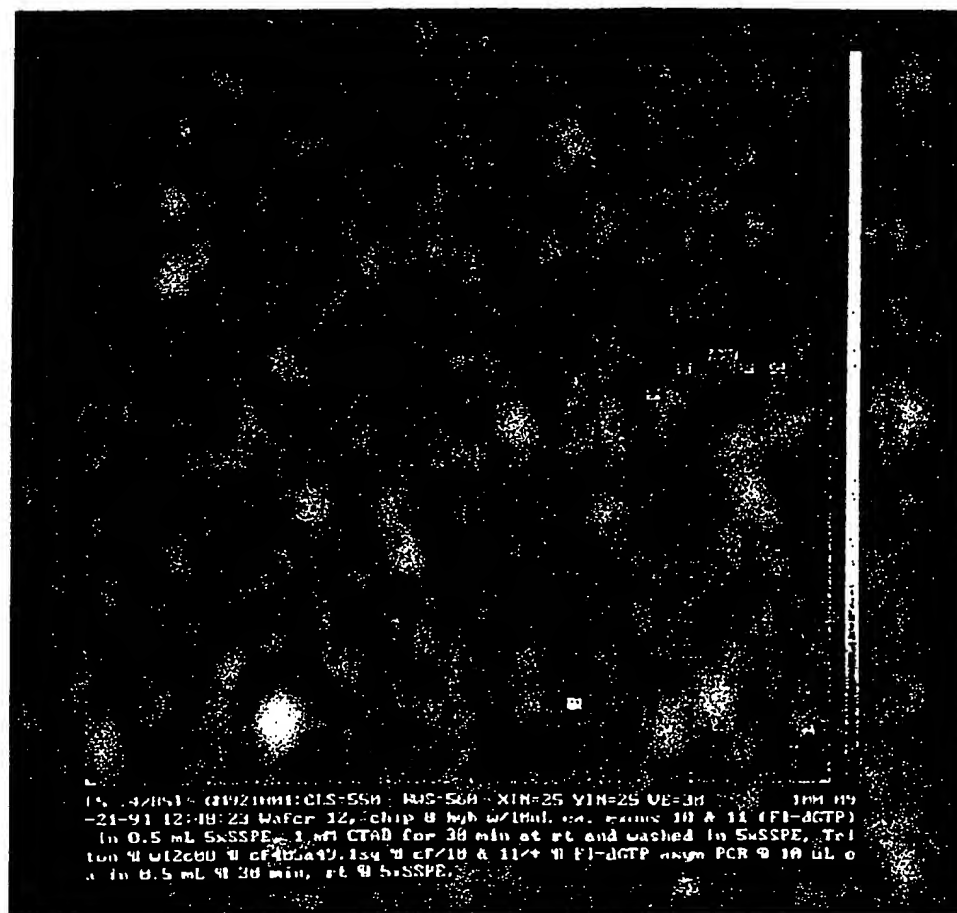


Fig. 25
Page 1 of 2

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B

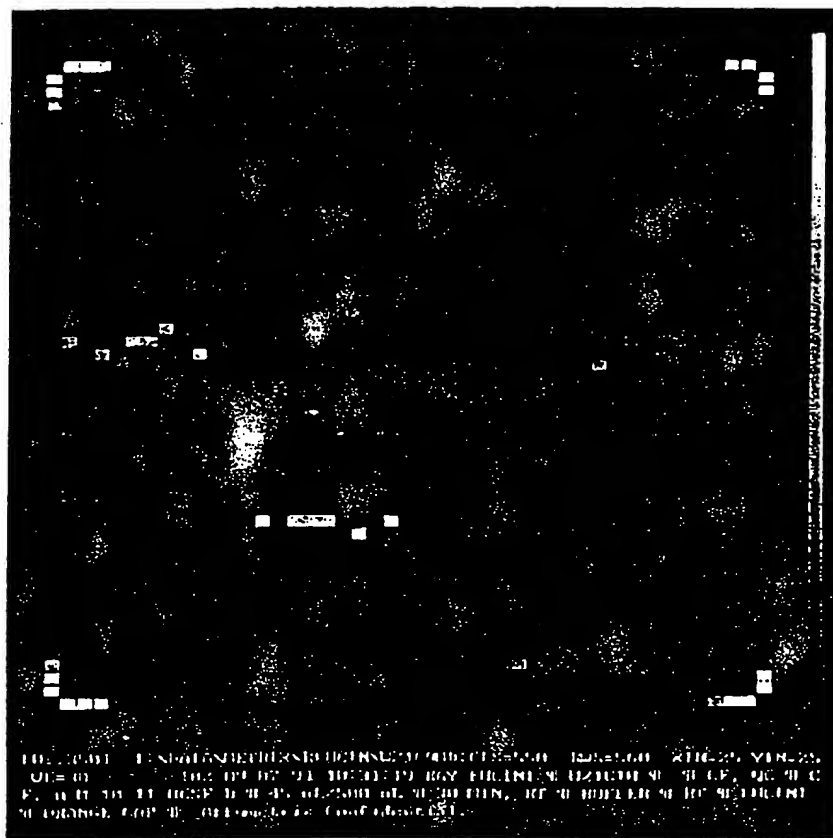


Fig. 25
Page 2 of 2

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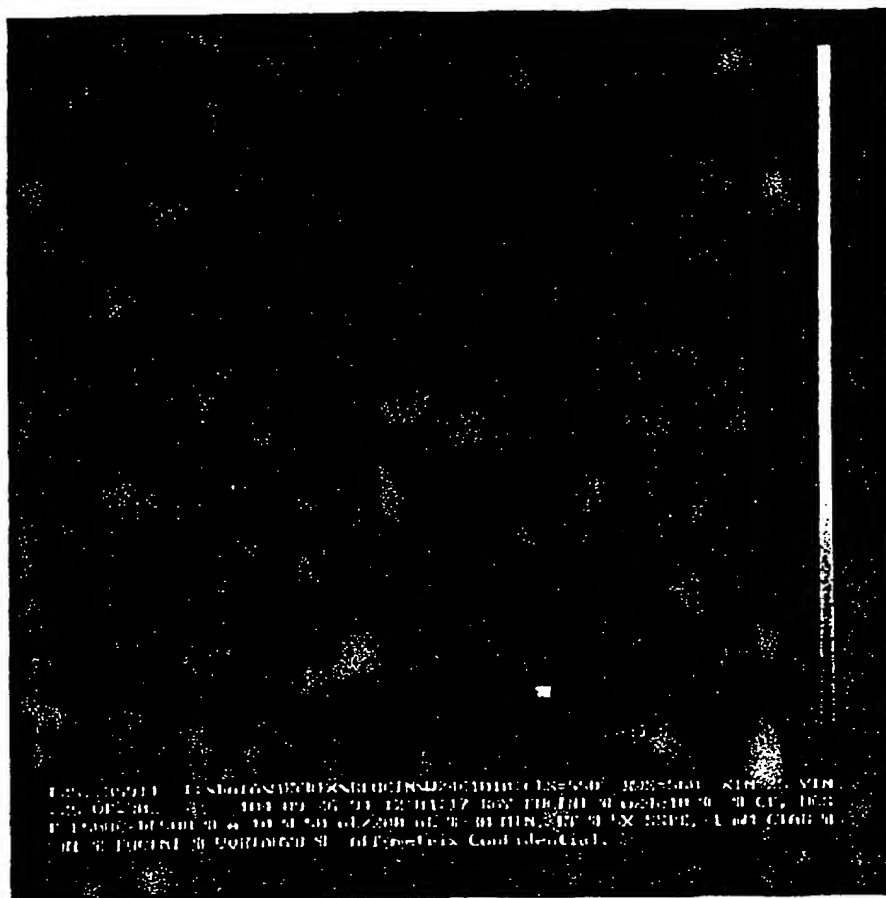


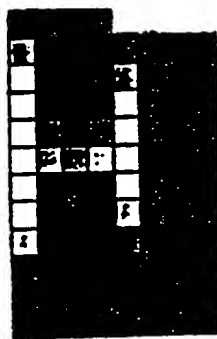
Fig. 26

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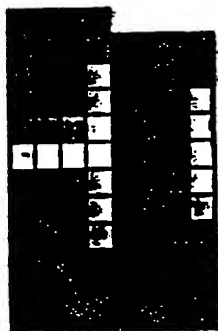
Detection of 12-mer One-Base Substitution P53 Targets

Fig. 29

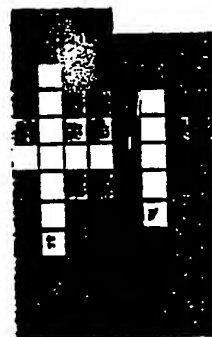
WT ("G" Substitution)
Target 12-mer



"A" Substitution 12-mer Target



"T" Substitution Target 12-mer

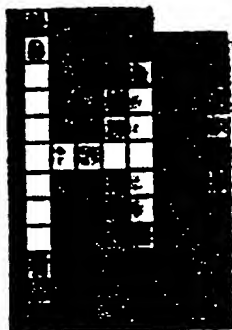


"C" Substitution Target 12-mer



Fig. 31

4:1 Mixture of WT and
"A" Substitution 12-mer
Targets



Figs. 29 and 31

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P53 EXON 6 CODON 192 REGION

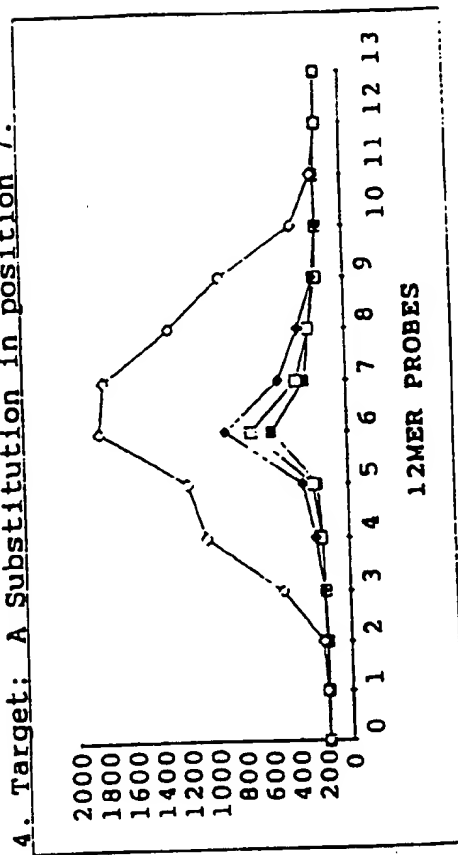
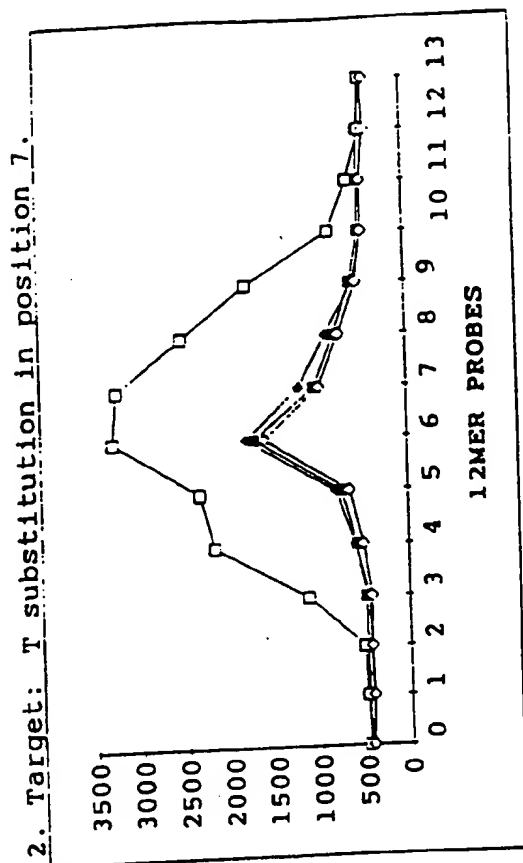
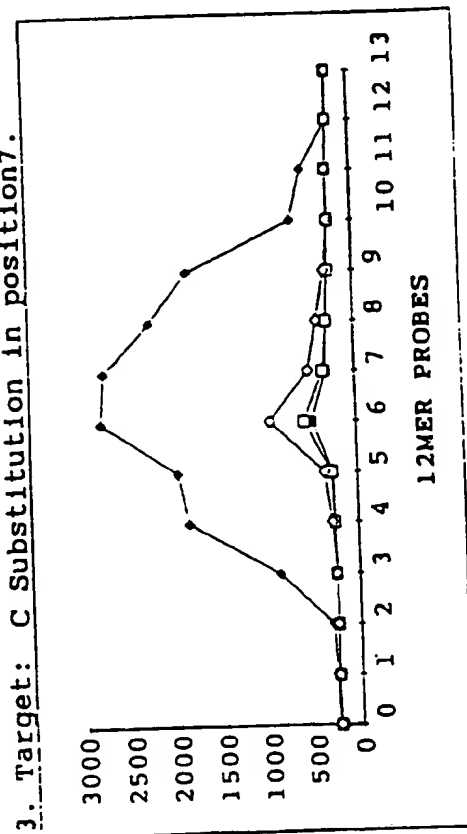
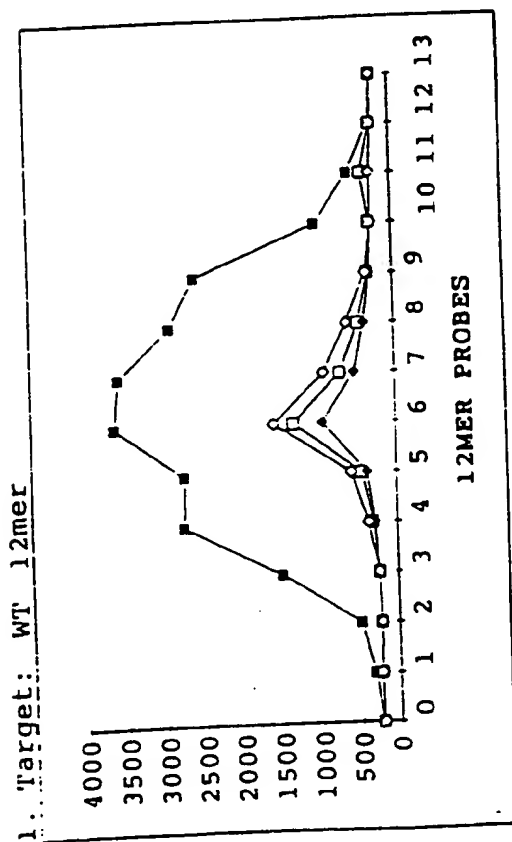


Fig. 30

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P53 EXON 6 CODON 192 REGION

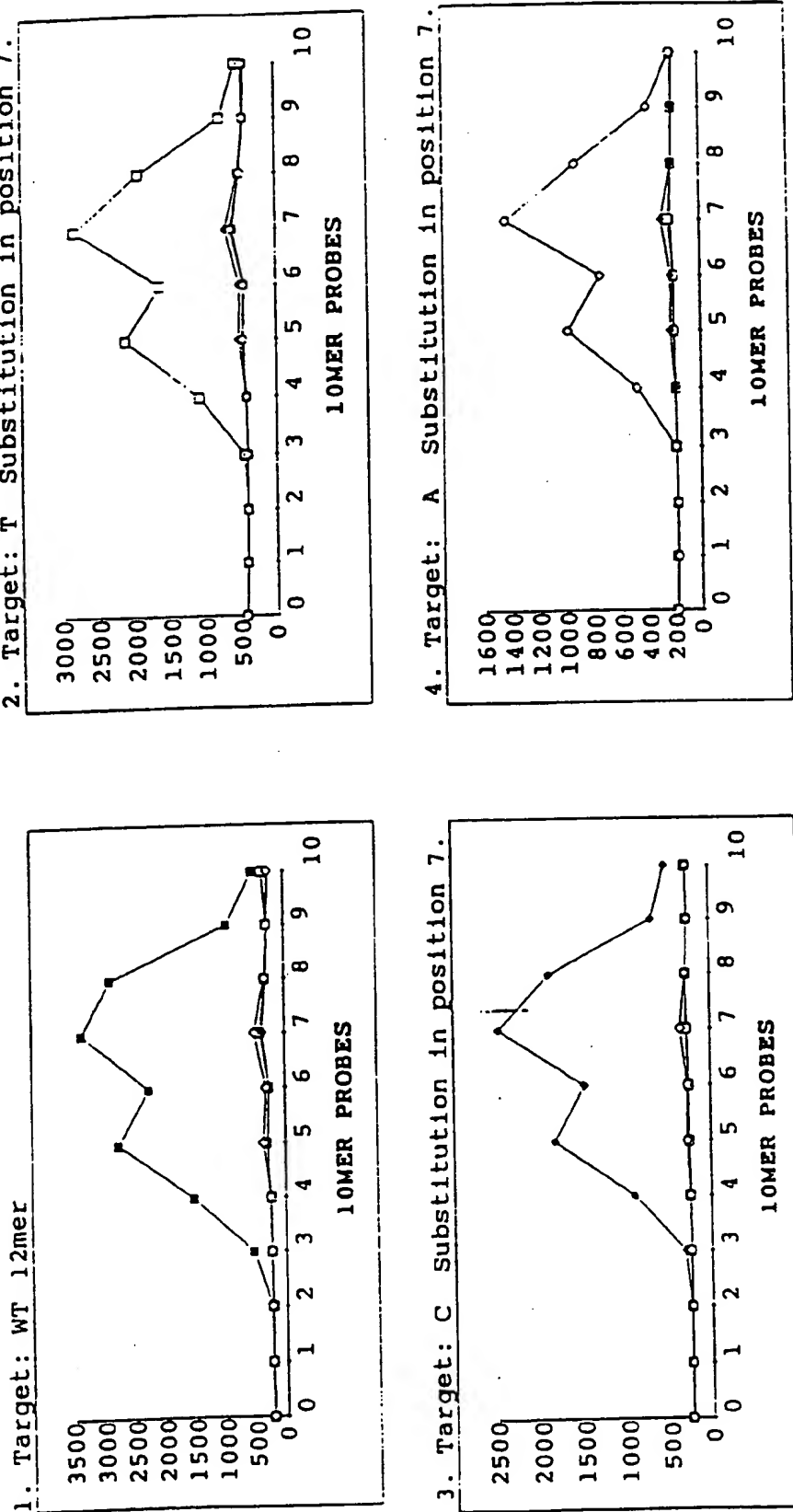


Fig. 32

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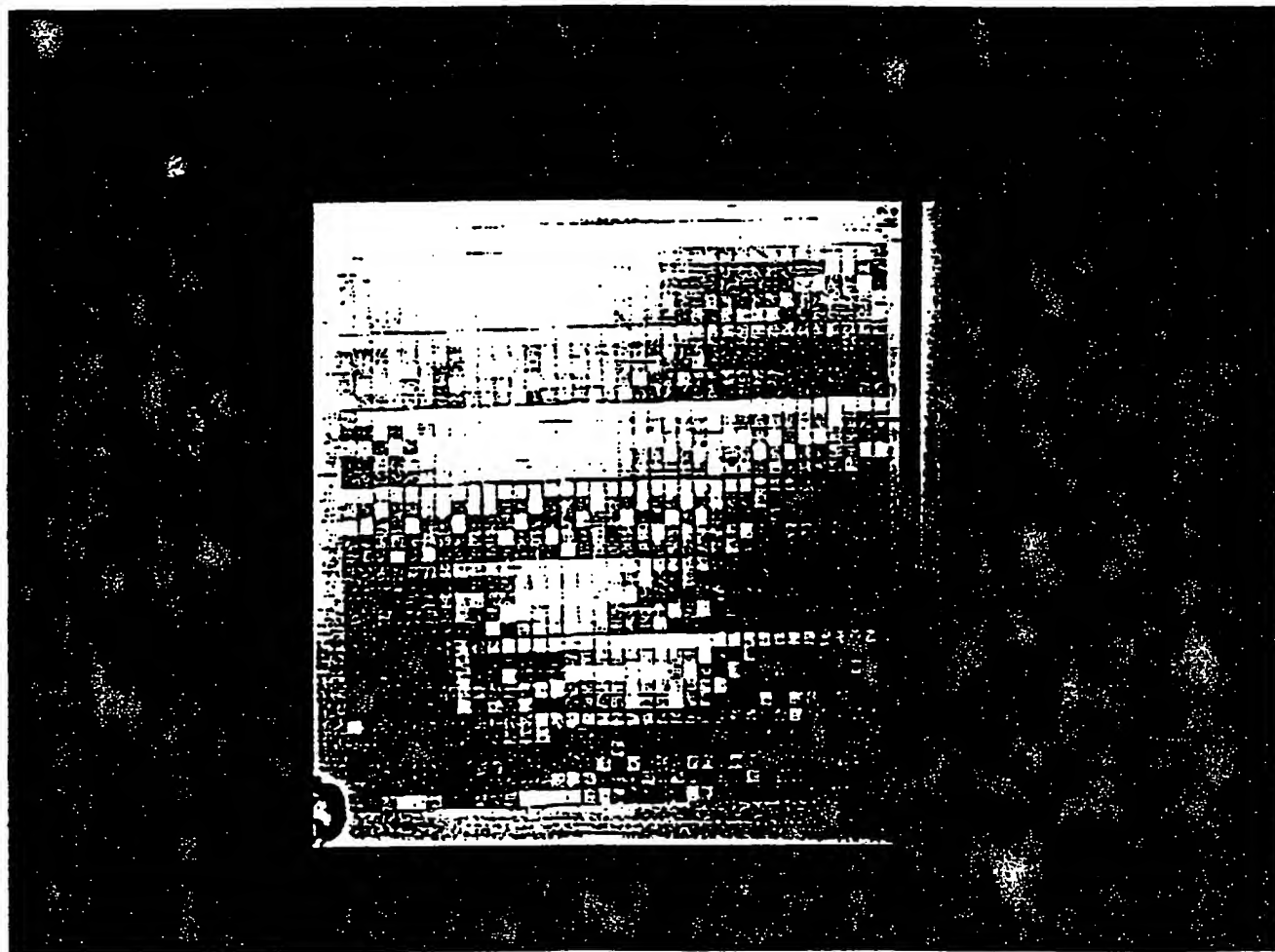


Fig. 33

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p53 Exon 5 Sequencing Key

WT	A	C	G	T	WT	A	C	G	T	WT	A	C	G	T	WT	A	C	G	T	WT	A	C	G	T	WT	A	C	G	T
T					G					C					C					T					G				
A					C					A					A					A					T				
C					T					C					C					C					G				
T					A					T					A					T					A				
G					C					G					C					G					C				
C					T					C					T					C					T				
A					G					A					A					A					G				
C					A					C					A					C					A				
T					C					T					C					T					C				
G					T					G					T					G					T				
C					A					C					A					C					A				
T					C					T					C					T					C				
A					G					A					A					A					G				
C					A					C					A					C					A				
T					C					T					C					T					C				
G					T					G					T					G					T				
C					A					C					A					C					A				
T					C					T					C					T					C				
A					G					A					A					A					G				
C					A					C					A					C					A				
T					C					T					C					T					C				
G					T					G					T					G					T				
C					A					C					A					C					A				
T					C					T					C					T					C				
A					G					A					A					A					G				
C					A					C					A					C					A				
T					C					T					C					T					C				
G					T					G					T					G					T				
C					A					C					A					C					A				
T					C					T					C					T					C				
A					G					A					A					A					G				
C					A					C					A					C					A				
T					C					T					C					T					C				
G					T					G					T					G					T				
C					A					C					A					C					A				
T					C					T					C					T					C				
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C					A					C					A					C					A				
T					C					T					C					T					C				
G					T					G					T					G					T				
C					A					C					A					C					A				
T					C					T					C					T					C				
A					G					A					A					A					G				
C					A					C					A					C					A				
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C					A					C					A					C					A				
T					C					T					C					T					C				
A					G					A					A					A					G				
C					A					C					A					C					A				
T					C					T					C					T					C				
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C					A					C					A					C					A				
T					C					T					C					T					C				
A					G					A					A					A					G				
C					A					C					A					C					A				

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THE HUMAN MITOCHONDRIAL GENOME

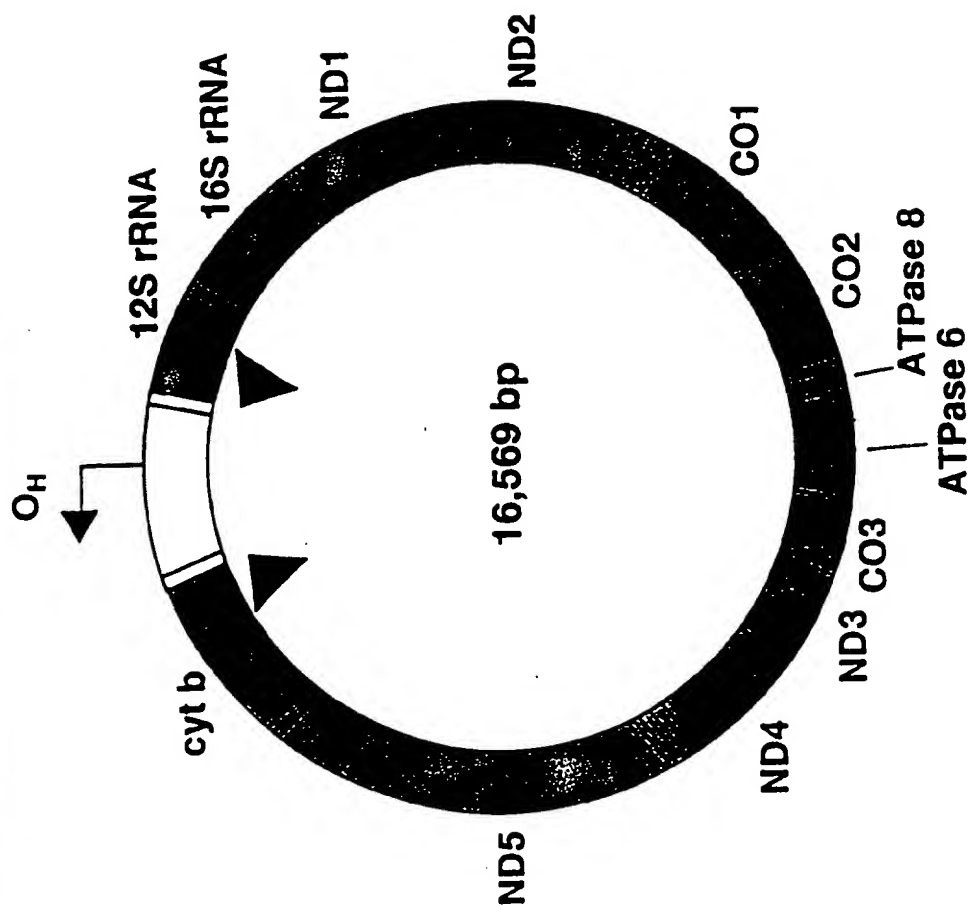
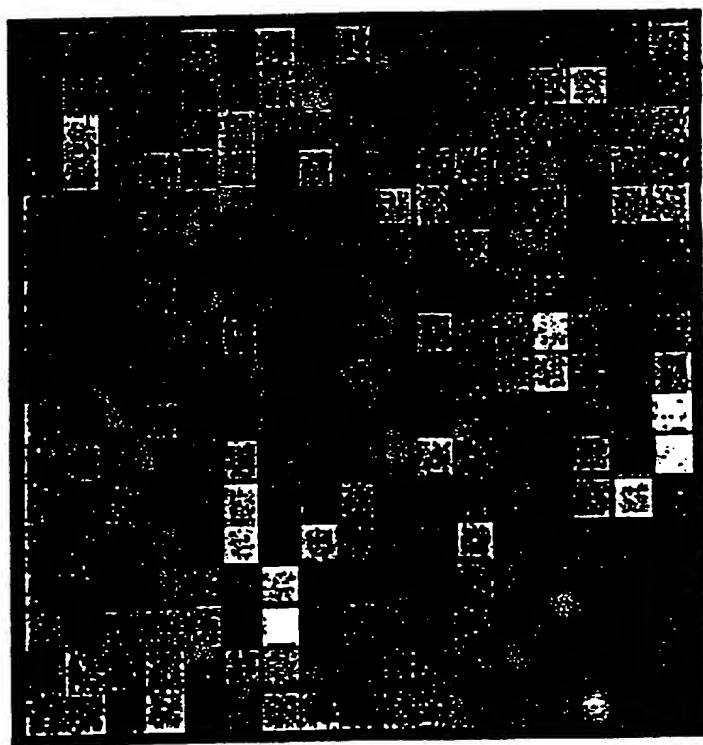


Fig. 35

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mt4

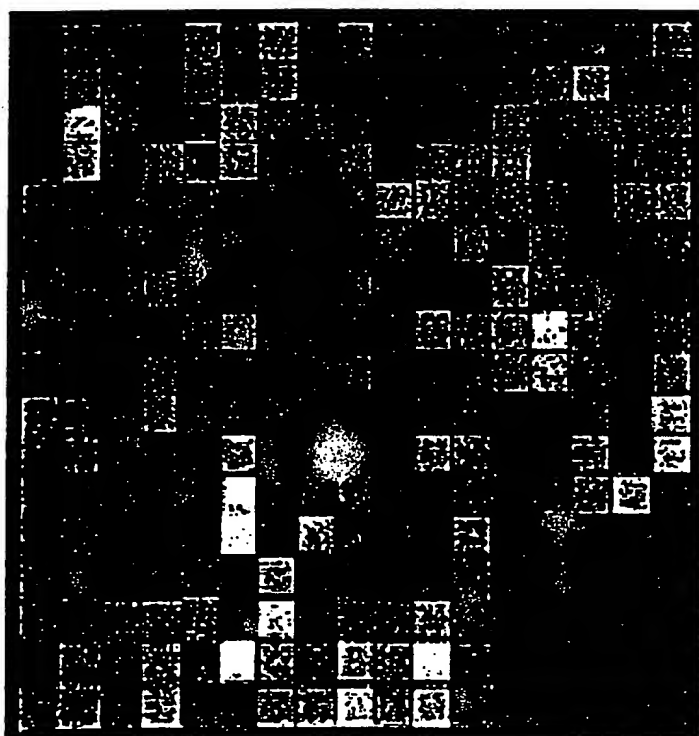


HYBRIDIZATION

Fig. 36

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mt5



HYBRIDIZATION

Fig. 37

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PREDICTED DIFFERENCE IMAGE

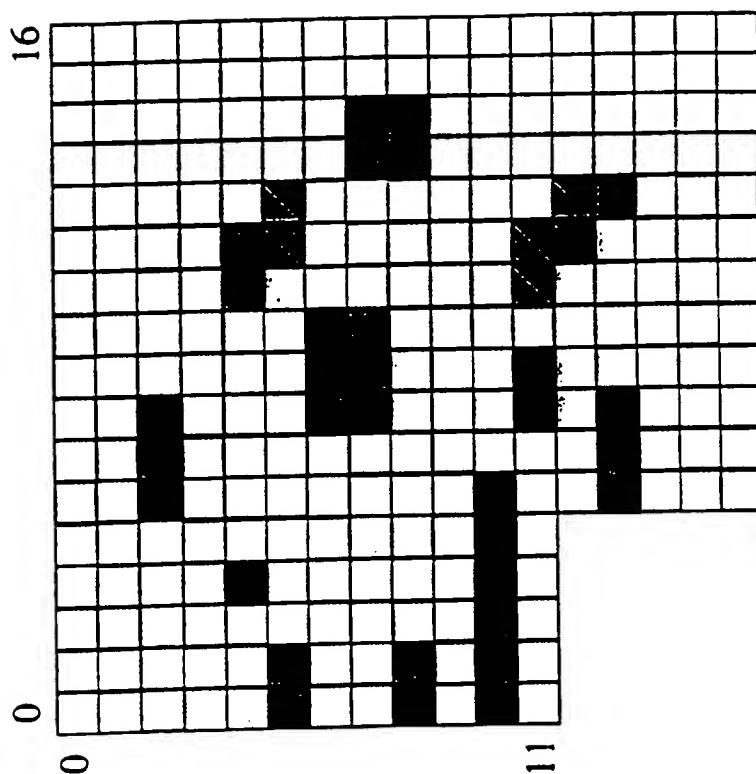


Fig. 38

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DIFFERENCE IMAGE

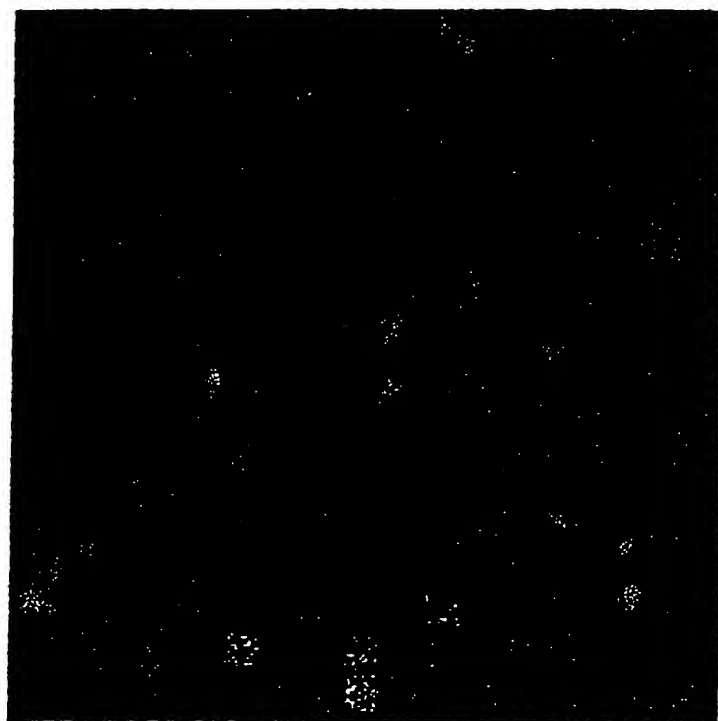


Fig. 39

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NORMALIZED INTENSITIES

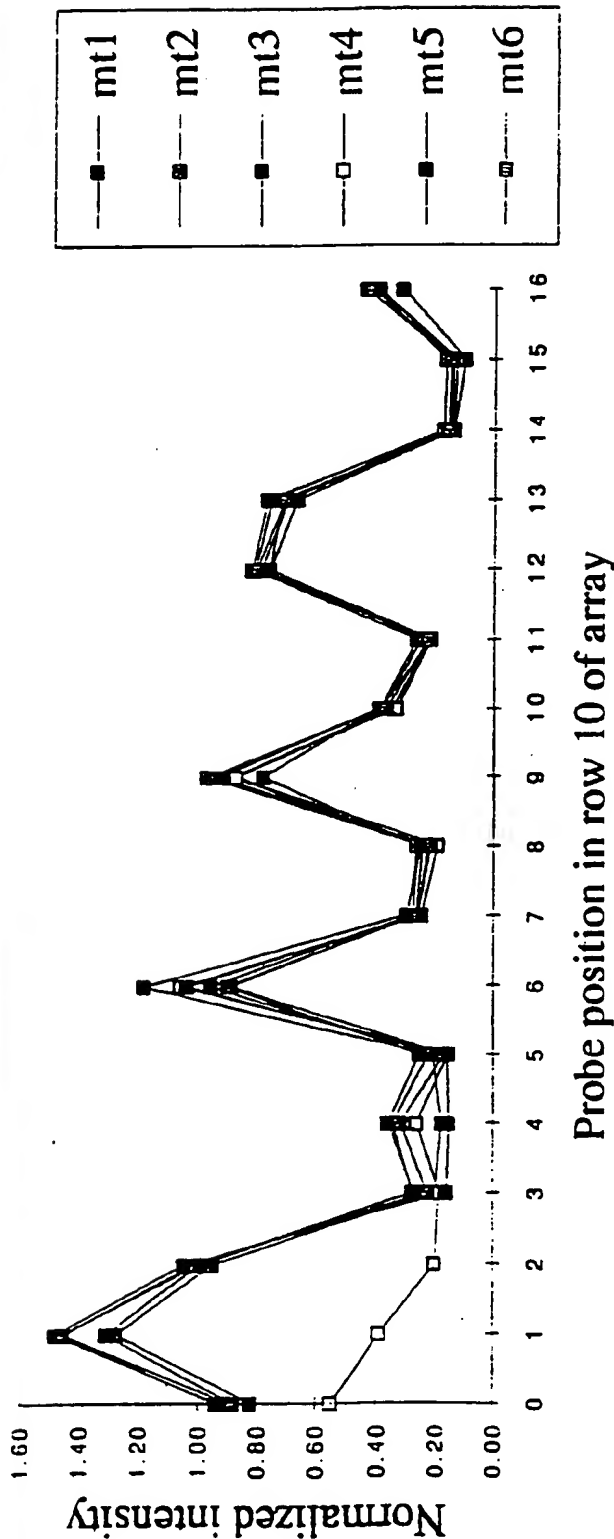


Fig. 40
Sheet 1 of 2

probe position	0	1	2	3	4	5
probe length	13	13	12	12	12	12
sample (mt1 -> 6)	4	4	4	2, 5	2, 5	2, 5
mismatch position from 3' of probe	12	5	3	12	7	2
base change	t -> a	t -> a	t -> a	t -> c	t -> c	t -> c

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NORMALIZED INTENSITIES

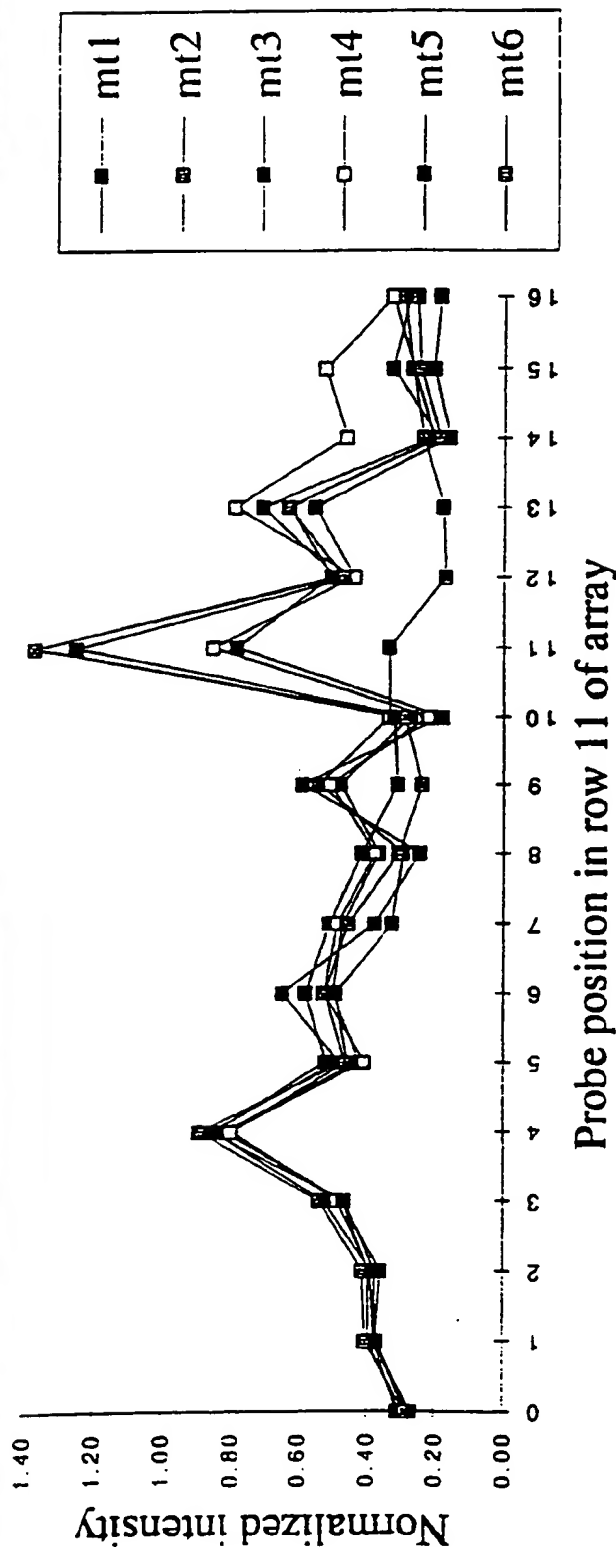


Fig. 40
Sheet 2 of 2

probe position	6	7	8	9	10	11	12	13
probe length	13	12	12	13	14	13	12	12
sample (mt1 -> 6)	2	2, 5	2, 5, 6	3, 6	3, 4, 5	2, 4, 5	2	2
mismatch position from 3' of probe	13	9, 10	3, 4 11	11, 5	4, 11, double	11, 3, double	6	3
base change	c -> t	c -> t	c -> t t -> c	t -> c	t -> c double	g -> a t -> c double	g -> a	g -> a

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DISCRIMINATION

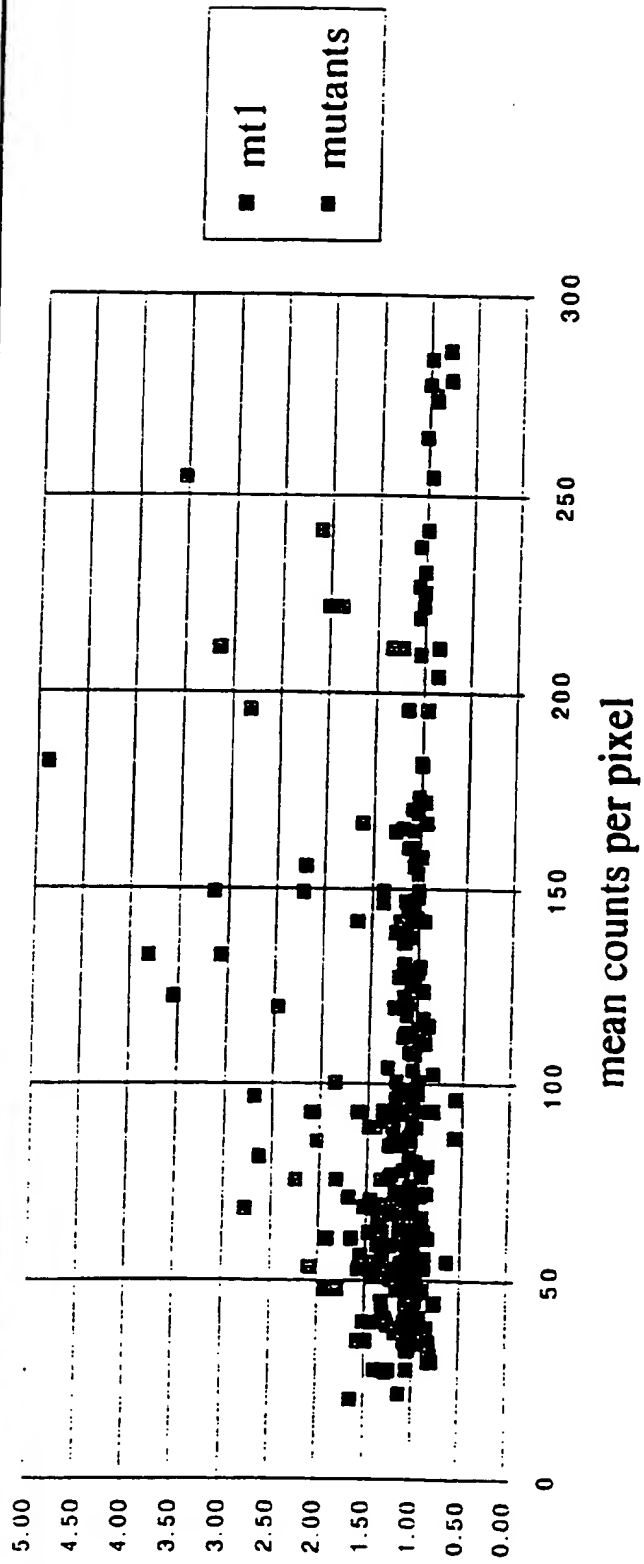


Fig. 41

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SEQUENCE & POSITION OF MUTATION

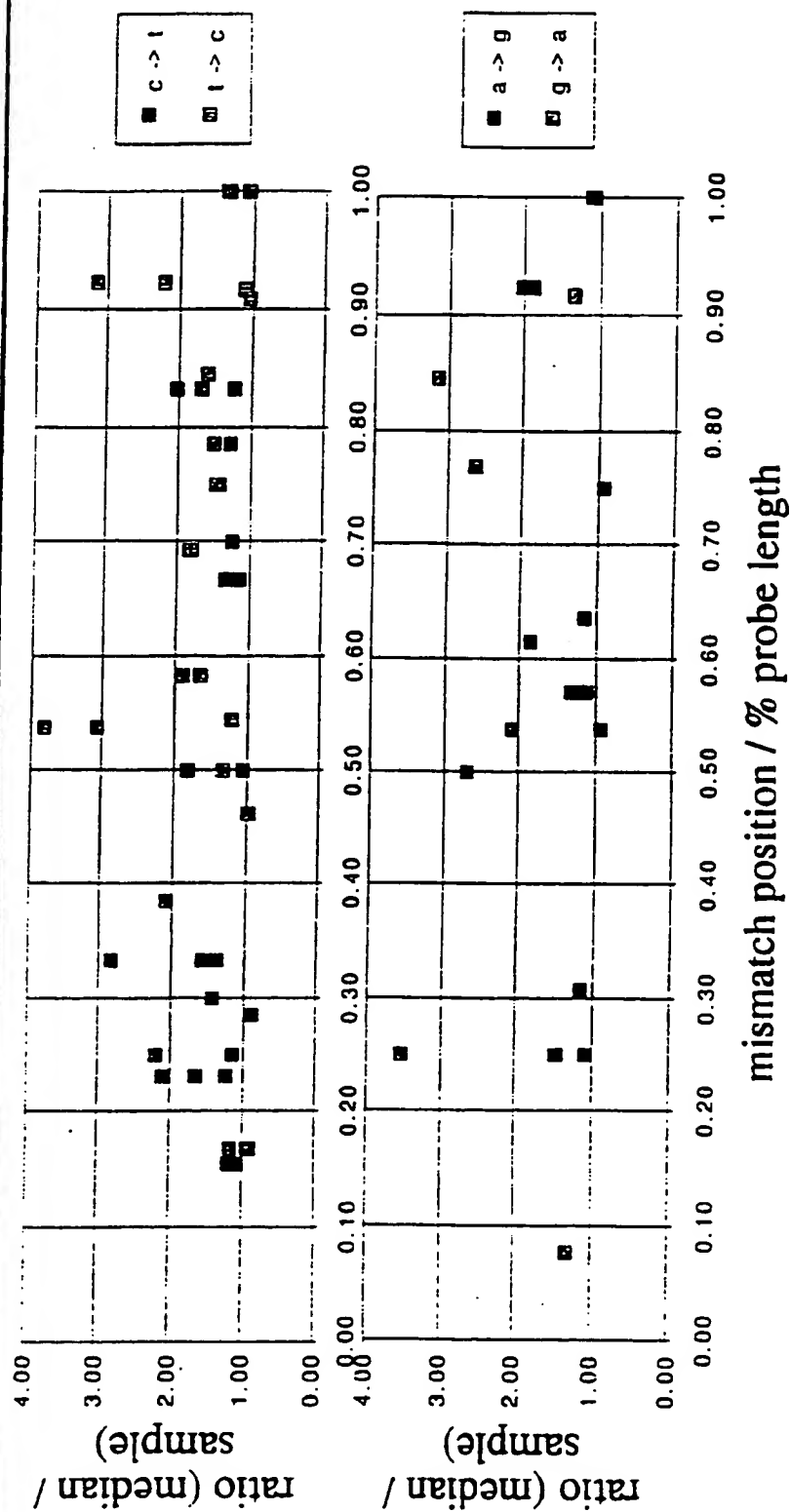


Fig. 42

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SEQUENCE

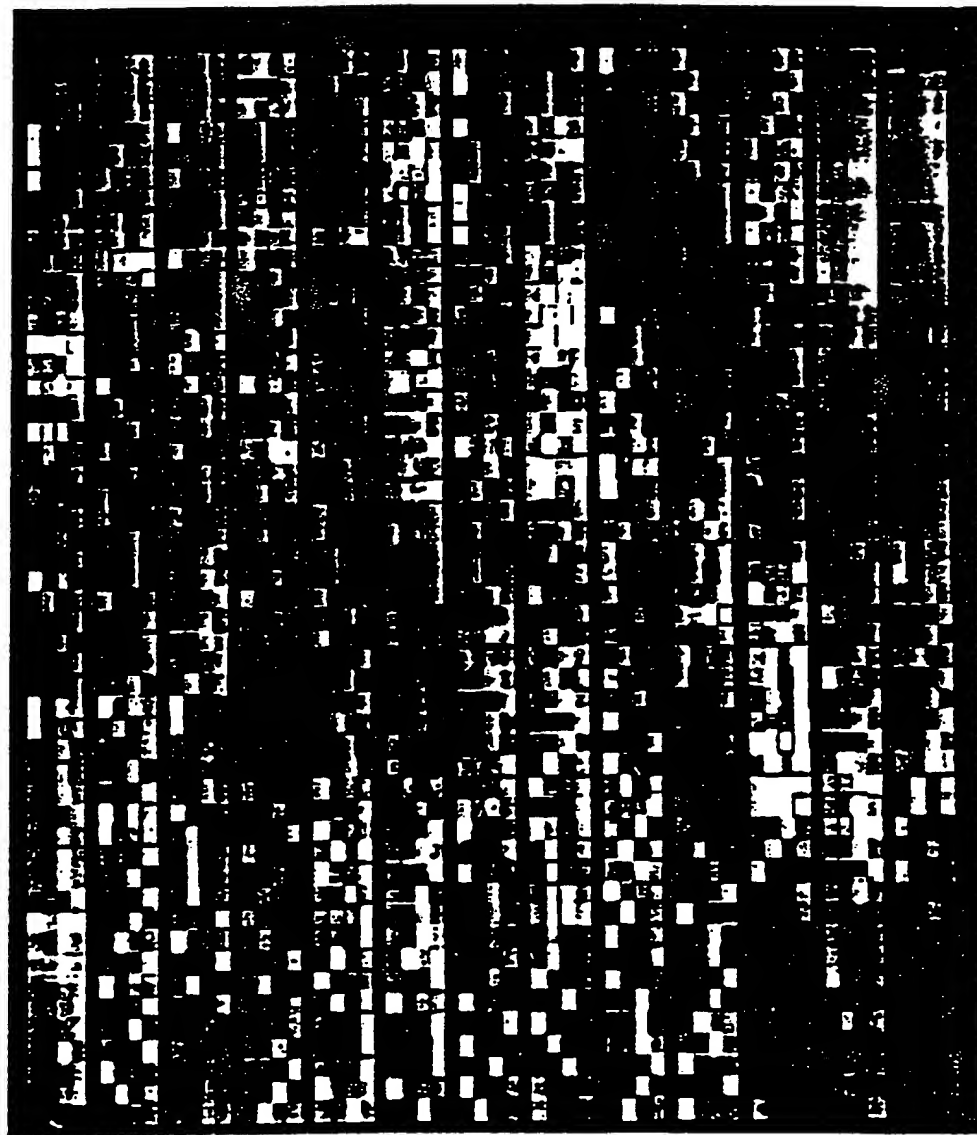
1 50
XaacaacctaaccaccccttaacagtagtacatagtaataaagccattttacX
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taatttaattaatgtcttgtaggacataataataacaattgaattgtctgcac
agcccActttccacacagacatcataaaaaaatttccaccaaaaccccc
XctcccccgcttctggccacagcacttaaacacatctTcttgccaaaccccX

Fig. 43

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Fig. 44

HYBRIDIZATION



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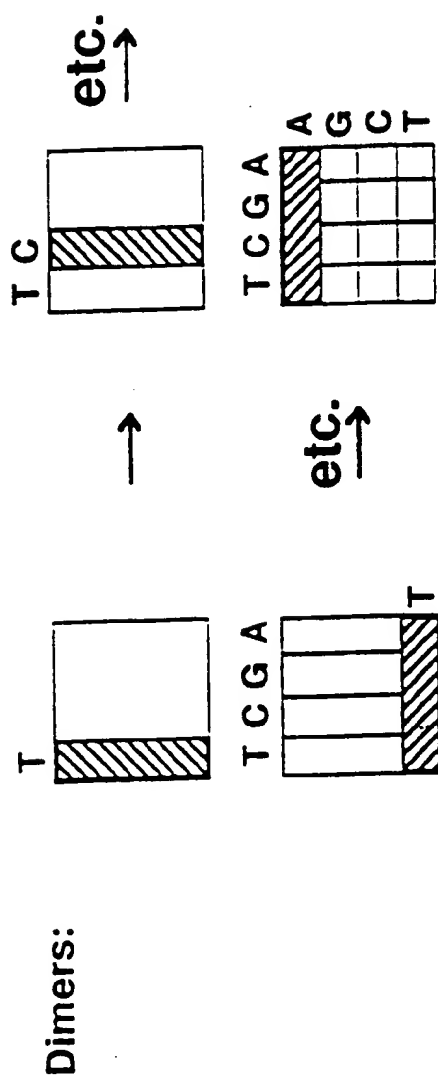
Position:	16519	152	263	344	
Change:	T->C	T->C	A->G	T->C	
Result:					T G C A

Fig. 45

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Nucleoside Combinatorials



in polynomial notation:
 $(T + C + A + G)^2 = \text{All Dimers}$

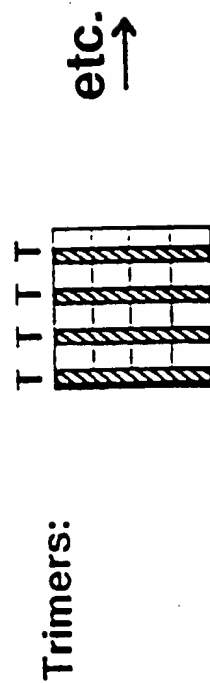
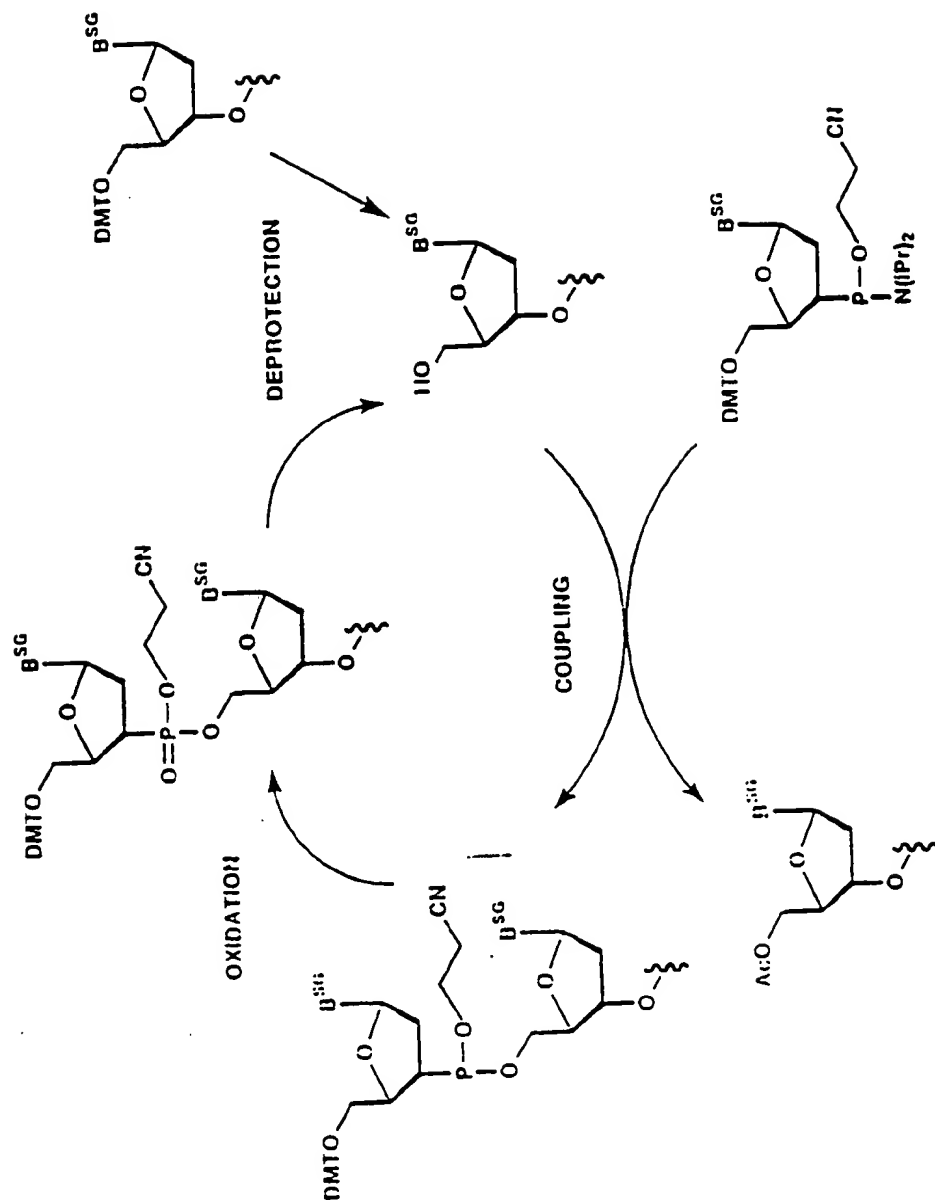


Fig. 47

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Fig. 48

Solid Phase DNA Synthesis



Nucleoside Buildingblocks

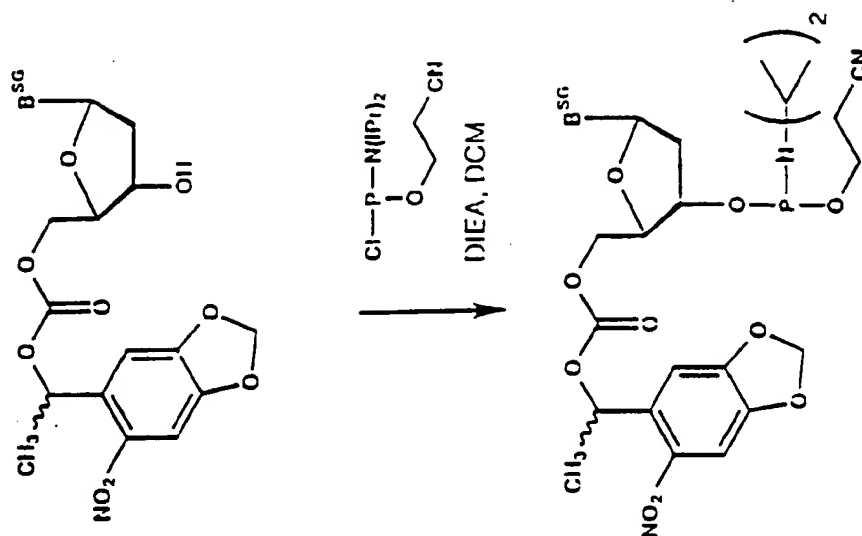
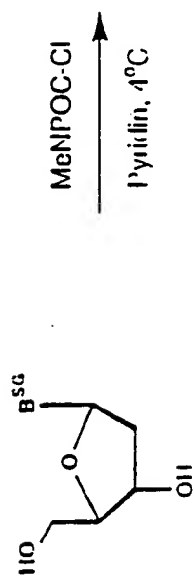
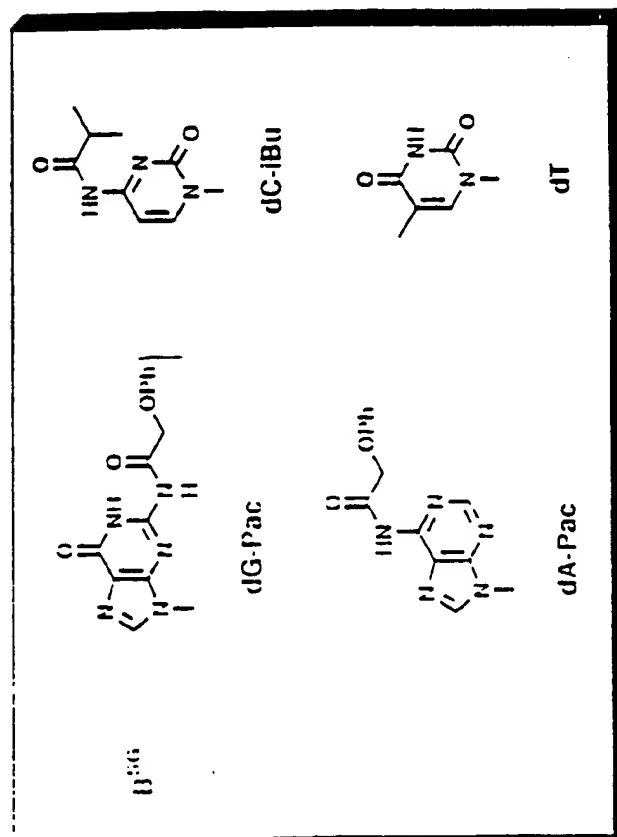


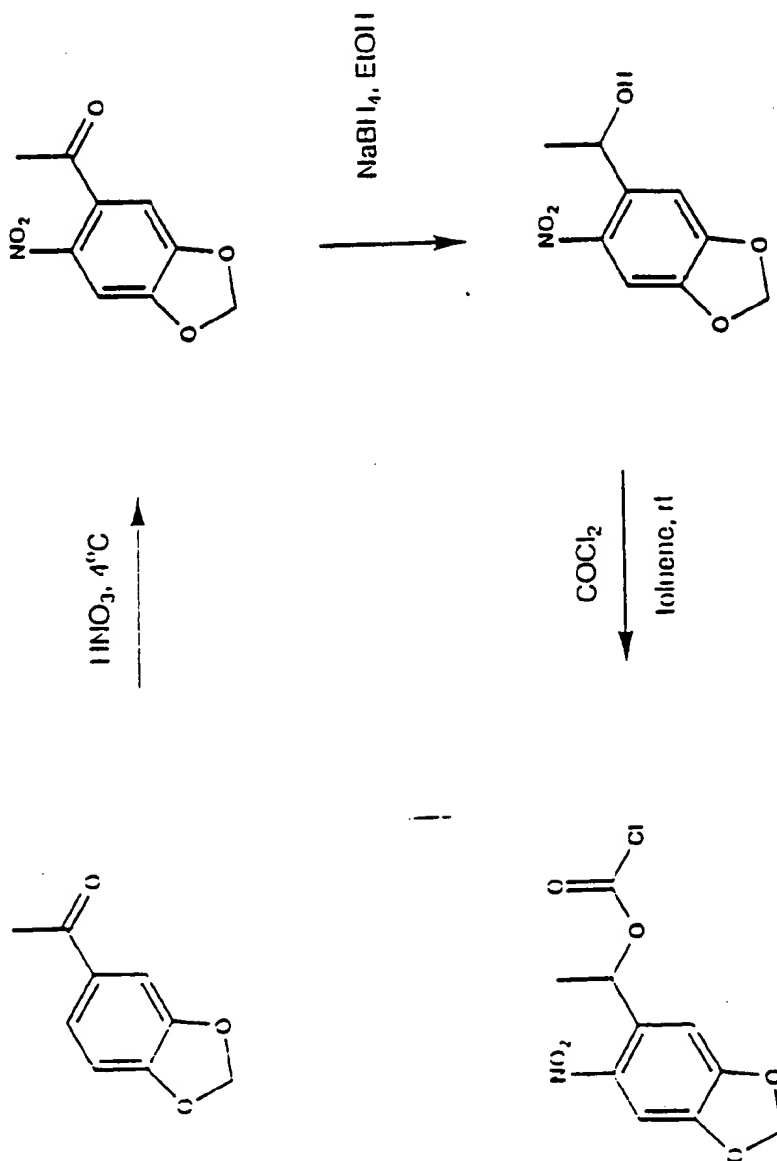
Fig. 49



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Fig. 50

MeNPOC-Cl



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Fig. 51

